

Juvenile Neuronal Ceroid Lipofuscinosis,
Childhood Dementia and Education

Juvenile Neuronal Ceroid Lipofuscinosis, Childhood Dementia and Education

Intervention, education and learning strategies
in a lifetime perspective

Edited by

*Stephen von Tetzchner, Bengt Elmerskog, Anne-Grethe Tøssebro
and Svein Rokne*

A step towards an extended curriculum
– Conclusions and recommendations from the
Erasmus+ sponsored project «JNCL and Education».

Published by:
SNØFUGL FORLAG
www.snøfugl.no

ISBN 978-82-7083-563-8

Design and cover:
IQ DESIGN
Janett Alice Olijnyk
www.iq-design.no

Heidi Tangen, Statped Midt, Norway,
has designed Figures 3.3, 16.4–16.7 and 16.10

Print:
INDEVELOP
Riga

Content

Foreword

Project Members and Authors

1 Introduction

*Stephen von Tetzchner, Bengt Elmerskog,
Anne-Grethe Tøssebro and Svein Rokne* 23

The NCL spectrum	23
The course of JNCL	24
Evidence-based practice	27
Education as a leading activity from childhood to emergent adulthood	28
JNCL and education	28
Registry	31
Overview	31
References	32

2 A Developmental Perspective on Juvenile Neuronal Ceroid Lipofuscinosis

Stephen von Tetzchner 35

The concept of development	35
Development may imply both progression and regression	39
The zones of proximal development and developmental maintenance	40
The role of planning in atypical development	42
References	43

3	Medical Aspects of Juvenile Neuronal Ceroid Lipofuscinosis <i>Alfried Kohlschütter, Jonathan D. Cooper, Sara E. Mole, John R. Østergaard and Angela Schulz</i>	45
	Definition of JNCL	45
	Genetic causes of JNCL	46
	Disease mechanisms in JNCL	47
	Diagnosis of JNCL	49
	Medical treatment of JNCL	51
	Common problems during disease progression	51
	Concluding comments	60
	References	60
4	Development and Loss of Vision <i>Bengt Elmerskog and Stephen von Tetzchner</i>	63
	Visual impairment	63
	Consequences of visual impairment	64
	Visual impairment in individuals with JNCL	68
	Conclusions	71
	References	72
5	Dementia in Adulthood and Childhood <i>Per Kristian Haugen, Stephen von Tetzchner, Judith D. Oxley and Bengt Elmerskog</i>	75
	Dementia	76
	Attention	78
	Memory	79
	Executive functions	82
	Dementia in JNCL	83
	Dementia and practice related to JNCL	87
	References	89
6	Language Development and Attrition in Juvenile Neuronal Ceroid Lipofuscinosis <i>Anne-Grethe Tøssebro and Stephen von Tetzchner</i>	93
	Language comprehension and production	93
	Language development in children with JNCL	94

	Language attrition in JNCL	95
	Observations and assessments	101
	Consequences of language loss	104
	References	105
7	Motor Development and Loss in Individuals with Juvenile Neuronal Ceroid Lipofuscinosis <i>Bengt Elmerskog and Riikka Hokkanen</i>	109
	Motor functions and impairment	109
	Decline of motor function and activities in individuals with JNCL	111
	Visual impairment	113
	Effect of motor and visual impairments on activities	114
	Physical activity, cognition and childhood dementia	117
	Dementia, physical activity and JNCL	119
	Conclusions	119
	References	120
8	Ethical Issues Related to Working with Individuals with Juvenile Neuronal Ceroid Lipofuscinosis <i>Stephen von Tetzchner and Finn Hesselberg</i>	125
	Ethical reflection and professional practice	126
	Understanding the lifeworlds of people with disabilities	130
	Disability, ethics and human rights	131
	Ideal and pragmatic services	133
	Concluding remarks	134
	References	135
9	Organization of Education and Counseling <i>Evelyn de Lorent, Cari Mannion, Marja-Leena Forssas, Tarja Eskonen, Anne Hagedorn Hamann, Susan Fugger, Merete Staureby, Bengt Elmerskog and Finn Hesselberg</i>	139
	Germany: Educating children and adolescents with JNCL in Hamburg	139
	Scotland: Aspects of the Scottish Education System	149
	Finland: Valteri Centre for Learning and Consulting	153
	Denmark: The Danish NCL Team	155
	Norway: The structure and function of services	160
	References	164

10	Assessment and Evaluation	
	<i>Stephen von Tetzchner, Anne-Grethe Tøssebro, Heather Adams, Barbara Cole and Bengt Elmerskog</i>	165
	Assessment strategies and processes and JNCL	166
	The Educational Development Observation Tool (EDO)	171
	References	176
11	Planning Education and Intervention for Individuals with Juvenile Neuronal Ceroid Lipofuscinosis (JNCL)	
	<i>Bengt Elmerskog, Stephen von Tetzchner, Anne-Grethe Tøssebro and Finn Hesselberg</i>	179
	Person-centered planning	180
	Habilitation Plans	181
	Individual Education Plans	184
	Reflections on educational planning	187
	Principles for making plans for persons with JNCL	191
	Core issues in educational planning	194
	A planning tool for JNCL	197
	Conclusions	197
	References	198
12	Early and Later Educational Intervention	
	<i>Anne-Grethe Tøssebro, Stephen von Tetzchner, Bengt Elmerskog and Aine Murphy</i>	201
	Adaptive education and special needs education	203
	Three educational phases	204
	Mathematics education and visual impairment	214
	Teaching mathematics to students with JNCL	216
	Drama in the education of students with JNCL	222
	References	226

13	Communication and Language in Education and Intervention for Children and Adults with Juvenile Neuronal Ceroid Lipofuscinosis <i>Anne-Grethe Tøssebro, Stephen von Tetzchner, Anna Christal, Deirdre McElrone, Mary Siggs, Judith D. Oxley and June Cameron</i>	231
	Interventions to strengthen and maintain intelligible speech	231
	Augmentative and alternative communication (AAC)	233
	AAC and JNCL	236
	Manual signs	239
	Factors of importance for AAC intervention	243
	Some useful tools to promote successful communication	247
	Prioritizing communication	254
	The need for future research	254
	References	255
14	Teaching Reading and Writing to Students with Juvenile Neuronal Ceroid Lipofuscinosis <i>Anne-Grethe Tøssebro, Anna Christal, Deirdre McElrone, June Cameron, Marja Leena Forssas and Tarja Eskonen</i>	257
	Teaching of literacy	257
	Modes of reading	258
	Reading and writing for children with JNCL	263
	Braille education for students with JNCL	268
	Braille literacy and impact on life	269
	Strategies in teaching reading and writing skills	270
	Creative writing and fan fiction	277
	Three creative writers with JNCL	279
	Braille writing from a parent's perspective	280
	Giving priority to literacy	285
	References	287

15	Physical Education and Activity for Individuals with Juvenile Neuronal Ceroid Lipofuscinosis	
	<i>Bengt Elmerskog, Svein Rokne, Bjørg Stenersen, Rita Jeremiassen and Riikka Hokkanen</i>	289
	Physical activity and JNCL	289
	Some factors related to physical activity in individuals with JNCL	295
	Physical activities for individuals with JNCL	297
	Two stories	304
	References	309
16	Coping with Everyday Life	
	<i>Bengt Elmerskog, Stephen von Tetzchner, Marja-Leena Forssas and Tarja Eskonen</i>	311
	Independence and interdependence	312
	Education for coping with everyday life	314
	Decline in independence	318
	Step-by-step support of coping with daily life	322
	Suggestions from parents and staff	338
	References	340
17	Music Education and Music in Education	
	<i>Adam Ockelford and Rebecca Atkinson</i>	343
	Main results from the survey	343
	The potential function of music for children and young people with JNCL	346
	References	358
18	Musical Action to Support Children and Young People with Juvenile Neuronal Ceroid Lipofuscinosis	
	<i>Jochen Lippe Holstein</i>	359
	Musical action	359
	Nurture despite degenerative disease	362
	Music as an occasion and goal for common action	363
	How to make music together	365
	Examples of musical action	366
	Conclusions	370
	References	371

19	Technology in Education	
	<i>Mohammed Beghdadi</i>	373
	Aims, possibilities and challenges in using technology	374
	Compensation through use of audio devices in teaching	376
	Compensation through use of speech recognition	378
	Assistive devices to support inclusion	378
	Assistive devices to promote motivation	379
	Some tools that may be useful in education of students with JNCL	380
	Tools for supporting communication	381
	Electronic tools for daily living	393
	Recommended computer equipment and other aids	396
	References	397
20	Quiz Games for Building and Maintaining Knowledge	
	<i>Mohammed Beghdadi and Bengt Elmerskog</i>	399
	Quiz in education	399
	Learning facts and making personal reflections	400
	Refreshment and consolidation of learning	401
	Quizzes and teaching	401
	Quizzes for people with dementia	402
	Quiz use among children and young people with JNCL	403
	Quiz Tool for persons with JNCL	405
	Conclusions	410
	References	410
21	Games	
	<i>Bengt Elmerskog, Mohammed Beghdadi, Svein Rokne, Anna Christal, Deirdre McElrone, Judith D. Oxley and June Cameron</i>	413
	Experiences of parents and staff	414
	Modifications and engagement	417
	A variety of games	418
	Conclusion	429
	References	430

22	The Social Life of Children and Adolescents with Juvenile Neuronal Ceroid Lipofuscinosis: School and Participation <i>Stephen von Tetzchner, Anne Margrethe Andersen, Anne Hagedorn Hamann and Susan Fugger</i>	431
	Vertical and horizontal relationships	431
	Activities, peer relations and JNCL	432
	Social adaptation and intervention	440
	The Danish summer school	442
	References	448
23	Transition to Adult Living <i>Svein Rokne and Bengt Elmerskog</i>	451
	Focus on participation in activities	452
	From adolescence to adulthood	458
	Findings from the JNCL project	459
	The preparatory phase	461
	Life as an adult	466
	Conclusions	473
	References	473
24	The Diagnostic Process and Parental Needs and Support <i>Svein Rokne, Bengt Elmerskog, Merete Staureby, Susan Fugger, Anne Hamann and Susanne Joensen</i>	475
	A typical story	475
	Requirements for guidelines for the diagnostic process	477
	Danish guidelines when a child is found to have JNCL	478
	How are parents supported?	480
	The immediate support phase	482
	References	483
25	Being the Brother or Sister of a Child with Juvenile Neuronal Ceroid Lipofuscinosis <i>Minna Sarola, Egil Rian, Linn Sophie Rian, Charlotte Holmen and Ida Holmen</i>	485
	Six families' views on sibling relations	485
	Reflections of a sister and a mother	490
	Perspectives on siblings	495
	References	503

26	Family Organizations and Advocacy Groups <i>Barbara Cole, Svein Rokne, Iris Dyck, Margie Frazer, Susan Fugger, Anne Hagedorn Hamann, Susanne Joensen, Carrie Mannion, Carsten Munkholm, Sabine Pilgram, Maria Liisa Punkari and Merete Staureby</i>	505
	Norsk Spielmeier-Vogt Forening (The Norwegian NCL Family Association)	506
	NCL Danmark (The Danish NCL Family Association)	507
	NCL-Gruppe Deutschland e.V. (The German NCL Family Association)	510
	Batten Disease Family Association (BDFA)	512
	The Batten Disease Support and Research Association (BDSRA)	515
	The Finnish NCL Family Association	516
	Links to the associations	517
27	Behavioral and Emotional Reactions to a Difficult Life Situation <i>Stephen von Tetzchner, Jochen Lippe Holstein and Per Kristian Haugen</i>	519
	Emotional and behavioral reactions and JNCL	519
	Vision loss, dementia and behavioral and emotional reactions	524
	Concluding comments	530
	References	532
	Afterword	535
	Appendix A Methods <i>Bengt Elmerskog, Stephen von Tetzchner, Anne-Grethe Tøssebro and Svein Rokne</i>	537
	Participants	537
	Surveys and interviews	540
	Procedures	542
	Analysis	542
	Ethics	542
	References	542
	Appendix B Resources	543

Foreword

This book presents broad information about education and non-medical interventions for children, adolescents and adults with juvenile neuronal ceroid lipofuscinosis (JNCL). This is a serious disease and most research on JNCL and other NCL diseases concerns medical aspects. The low prevalence of JNCL may have contributed to the limited non-medical research. Educational guidelines for practice and coherent information about the typical course and variation in the development of JNCL have been lacking. Educational practice tends to be based on experience obtained from teaching individual students or from input provided by small support and advocacy organizations. Owing to language and cultural barriers and limited resources, the individual knowledge bases have not been pooled successfully to others who work with persons with JNCL, and written documentation hardly exists. Dedicated teachers and other professionals have lacked both research-based and systematic experience-based knowledge about educational and other non-medical interventions that can support development, learning and skill maintenance in individuals with JNCL. There is thus a need for more research to determine innovative and varied approaches within educational and other services to meet the challenges of the disease. Also needed is a strategy to pool current knowledge and disseminate so that all stakeholders can draw on it.

The present book thus builds on existing literature and adds to it by reporting findings from an international project. This project draws on findings from seven countries. It includes findings from a comprehensive survey and interviews with a relatively large number of families and professionals working with this group (e.g., teachers, special educators and residential staff). The project also includes the development of educational tools and strategies and small trials. The authors include researchers and practitioners with many years of experience with working for persons with JNCL, as well as family members. The book gives guidelines for practice and suggestions based on information about the typical course and the considerable variation that exists in learning and development within this group.

The book is intended for teachers, special educators and other professionals working with children, adolescents and adults with JNCL, as well as for families with a member who has JNCL. The book addresses educational and other interventions for individuals who show visual and cognitive decline with onset in childhood, and it may be useful also for families and professionals working with other diagnostic groups with similar problems.

Acknowledgement

This book is a part of the international project «Juvenile Neuronal Ceroid Lipofuscinosis (JNCL) and Education» (2014–2017) which was funded by the European Commission through Erasmus+ grant No. 2014-1-NO01-KA200-000388. In addition, the project was supported by participating organizations and volunteer work. Preparatory activities in 2013 and 2014 were funded by The Norwegian Directorate for Children, Youth and Family Affairs, The Norwegian Directorate for Education and Training and the NCL Family Association in Norway and Statped, Norway. Statped Midt in Norway has coordinated the project. The following institutions have been funded by the project:

Batten Disease Family Association (BDFA), Farnborough, UK
Bildungszentrum für Blinde und Sehbehinderte (BZBS), Hamburg, Germany
Norsk Spielmeyer-Vogt Forening, Norway
Oppimis-ja ohjauskeskus Valteri, Onerva, Finland
Royal Blind School, Edinburgh, Scotland
Statped Midt, Trondheim, Norway
WESC Foundation, Exeter, UK

The following organizations were members of the project but funded through other sources:

Batten Disease Support and Research Association (BDSRA), USA
Chiltern Music Therapy, UK
NCL Danmark, Denmark
Department of Psychology, University of Oslo, Norway
Finnish Federation of the Visually Impaired, Finland
LVR-Johanniterschule, Germany
NCL Gruppe Deutschland, Germany
New College Worcester, UK

University Medical Center Eppendorf, Germany
University of Rochester Medical Center, USA
University of Roehampton, UK
Vision Centre Refsnæs, Denmark

We want to thank the participating families and professionals who shared their knowledge and gave of their time. Our sincere thanks go to all organizations and individuals who have contributed to the project with insights, background material, relevant experiences and life stories which form the backbone of the present work.

The Editors
Oslo, Trondheim, Asker
January 2019

Project Members and Authors

Heather Adams
Department of Neurology
University of Rochester Medical Center
New York, USA
Email: heather_adams@urmc.rochester.edu

Anne M. Andersen
Vision Centre Refsnæs
Kalundborg, Denmark
Email: aman@regionsjaelland.dk

Rebecca Atkinson
Chiltern Music Therapy
Chesham, London, UK
Email: rebecca@chilternmusictherapy.co.uk

Heather Band
Batten Disease Family Association (BDFA)
209-211 City Road, London, UK
Email: heatherband@bdfa-uk.org.uk

Mohammed Beghdadi
Statped Midt
Trondheim, Norway
Email: mohammed.beghdadi@statped.no

Elaine Brackenridge
Royal Blind School
Edinburgh, Scotland, UK
Email:
elaine.brackenridge@royalblindschool.org.uk

Mark Braithwaite
WESC Foundation
Exeter, UK
Email: mbraithwaite@wescfoundation.ac.uk

June Cameron
Royal Blind School
Edinburgh, Scotland, UK
Email: june.cameron@royalblindschool.org.uk

Anna Christal
Royal Blind School
Edinburgh, Scotland, UK
Email: anna.christal@royalblindschool.org.uk

Barbara Cole
Batten Disease Family Association (BDFA)
209-211 City Road, London, UK
Email: barbaracole@bdfa-uk.org.uk

Jonathan D. Cooper
Washington University School of Medicine
St. Louis, USA
Email: cooperjd@wustl.edu

Tracy de Bernhardt Dunkin
WESC Foundation
Exeter, UK
Email: tracybd@wescfoundation.ac.uk

Evelin de Lorent
Bildungszentrum für Blinde und
Sehbehinderte (BZBS)
Hamburg, Germany
Email: evelin.delorent@bsb.hamburg.de

Iris Dyck
NCL-Gruppe Deutschland
Berlin, Germany
Email: irisdyck@gmx.de

Bengt Elmerskog
Statped Midt
Trondheim, Norway
Email: belmerskog@gmail.com

Tarja Eskonen
Centre for Learning and Consulting, Onerva
Jyväskylä, Finland
Email: tarja.eskonen@valteri.fi

Marja-Leena Forssas
Centre for Learning and Consulting, Onerva
Jyväskylä, Finland
Email: marja-leena.forssas@valteri.fi

Per Fosse
Statped Midt
Trondheim, Norway
Email: per.fosse@statped.no

Margie Frazier
Batten Disease Support and Research
Association (BDSRA)
Columbus, Ohio, USA
Email: mfrazier@bdsra.org

Susan Fugger
Vision Centre Refsnæs
Kalundborg, Denmark
Email: sufu@regionsjaelland.dk

Editha Gombault
Bildungszentrum für Blinde und
Sehbehinderte (BZBS)
Hamburg, Germany
Email: edithagombault@googlemail.com

Anne Hagedorn Hamann
Vision Centre Refsnæs
Kalundborg, Denmark
Email: anne.hhamann@gmail.com

Per Kristian Haugen
Norwegian National Advisory
Unit on Ageing and Health
Tønsberg, Norway
Email: perkristian.haugen@aldringoghelse.no

Finn Hesselberg
Røn, Valdres, Norway
Email: finn.hesselberg@gmail.com

Riikka Hokkanen
Valteri Center for Learning and Consulting,
Onerva
Jyväskylä, Finland
Email: riikka.hokkanen@valteri.fi

Charlotte Holmen
Kirkenær, Grue, Norway
charlotte.holmen@dinassistanse.no

Ida Holmen
Kirkenær, Grue, Norway
Email: idacholmen@gmail.com

Rita Jeremiassen
Gimse, Norway
Email: rita.jeremiassen@outlook.com

Susanne Joensen
Vision Centre Refsnæs
Kalundborg, Denmark
Email: sjoen@regionsjaelland.dk

Alfried Kohlschütter
Department of pediatrics,
University Medical Center
Hamburg-Eppendorf, Germany
Email: kohlschuetter@uke.uni-hamburg.de

Katja Lenzing
Bildungszentrum für Blinde und
Sehbehinderte (BZBS)
Hamburg, Germany
Email: kollmorgenm@aol.com

Jochen Lippe-Holstein
LVR-Johanniterschule
Duisburg, Germany
Email: jochen.lippe-holstein@gmx.de

Carrie Mannion
Royal Blind School
Edinburgh, Scotland, UK
Email: catchcari@yahoo.co.uk

Deirdre McElrone
Royal Blind School
Edinburgh, Scotland
Email: deirdre.mcelrone@royalblindschool.org.uk

Sara E. Mole
MRC Laboratory for Molecular Cell Biology
University College London, UK
Email: s.mole@ucl.ac.uk

Carsten Munkholm
NCL Danmark
(The Danish NCL Family Association)
Virum, Denmark
Email: cmunkholm@live.dk

Aine Murphy
Royal Blind School
Edinburgh, Scotland, UK
Email: aine.murphy@royalblindschool.org.uk

Adam Ockelford
Department of Education,
University of Roehampton
London, UK
Email: a.ockelford@roehampton.ac.uk

Judith D. Oxley
Department of Communicative Disorders
University of Louisiana at Lafayette, USA
Email: oxleyjd55@gmail.com

John Rosendahl Østergaard
Centre for Rare Diseases,
Aarhus University Hospital
Aarhus, Denmark
Email: john.oestergaard@skejby.rm.dk

Sabine Pilgram
NCL-Gruppe Deutschland
Berlin, Germany
Email: sabine.pilgram@gmx.de

Maria Liisa Punkari
Finnish Federation of the Visually Impaired
Helsinki, Finland
Email: marialiisapunkari@gmail.com

Egil Rian
Trondheim, Norway
Email: egrias@online.no

Linn Sophie Rian
Trondheim, Norway
Email: sophiegrana@gmail.com

Svein Rokne
Norsk Spielmeier-Vogt Forening
(The Norwegian NCL Family Association)
Asker, Norway
Email: srokne@online.no

Minna Sarola
The Norio Centre
Helsinki, Finland
Email: minna.sarola@rinnekoti.fi

Angela Schulz
Department of pediatrics,
University Medical Center
Hamburg-Eppendorf, Germany
Email: an.schulz@uke.uni-hamburg.de

Sian Shaw
New College Worcester
Worcester, UK
Email: sshaw@ncw.co.uk

Mary Siggs
Edinburgh, Scotland
Email: mary.siggs@gmail.com

Merete Staureby
NCL Danmark
(The Danish NCL Family Association)
Virum, Denmark
Email: familien@staureby.dk

Björg Stenersen
Habiliteringstjenesten i Hedmark,
Sykehuset Innlandet
Furnes, Norway
Email: bjorg.stenersen@sykehuset-innlandet.no

Anne-Grethe Tøssebro
Statped Midt
Trondheim, Norway
Email: annegrethe.tossebro@gmail.com

Stephen von Tetzchner
Department of Psychology,
University of Oslo
Oslo, Norway
Email: s.v.tetzchner@psykologi.uio.no

Andrea West
Batten Disease Family Association (BDFA)
Farnborough, London, UK
Email: andreawest@bdfa-uk.org.uk

1

Introduction

*Stephen von Tetzchner, Bengt Elmerskog,
Anne-Grethe Tøssebro and Svein Rokne*

The present book addresses learning and coping in individuals with juvenile neuronal ceroid lipofuscinosis (JNCL) from a life span developmental and educational perspective. A life span perspective examines how human abilities and skills grow and decline through development (Baltes & Baltes, 1990). The present book examines teaching and other forms of intervention for individuals with JNCL throughout life, including both early intervention and support in the period after they have left school. The education and intervention for individuals with JNCL include both early intervention and later adaptation, reflecting the growth and decline in skills and abilities in this group (see Chapter 12). The chapters present a variety of teaching tools and strategies designed to support learning and coping, maintenance of skills and abilities, and quality of life from childhood to adulthood. The vision of education and learning as lifelong processes emphasizes the need to choose goals that go beyond the classroom.

The NCL spectrum

Neuronal ceroid lipofuscinosis (NCL) is a group of inherited, progressive neurodegenerative diseases that affect cells in the brain, the retina, the heart and the skeletal muscles (see Chapter 3). The onset of symptomatology varies from infancy to adulthood, and they share the combined characteristics of retinopathy, dementia and epilepsy (Cooper, 2010; Kohlschütter & Schulz, 2009; Mole, Williams, & Goebel, 2011; Mole & Haltia, 2015; Rakheja & Bennett, 2018). Categorizations of the NCLs have varied through the years (Haltia & Goebel, 2013; Rider & Rider, 1999) and recent classifications include 9–14 genetically distinct disorders (Haltia & Goebel, 2013; Kohlschütter & Schulz, 2009; Rakheja & Bennett, 2018; Williams & Mole, 2012), and 360 different mutations (Kousi, Lehesjoki, & Mole, 2012).

This spectrum of diseases has several names. Neuronal ceroid lipofuscinosis is commonly known as Batten disease but there are also variants named after the genes they are associated with. Infantile Batten disease is also called CLN1 disease, late infantile Batten disease is called CLN2 disease, and variants of late infantile Batten disease may be CLN5, CLN6, CLN7 or CLN8 disease. The juvenile form (JNCL), also called the Spielmeyer-Vogt disease, is caused mainly by the CLN3 gene but also by mutations in other CLN genes (see Chapter 3). In the present book, the term juvenile neuronal ceroid lipofuscinosis (and its acronym JNCL) is used, as it is well established internationally. JNCL symptoms were first reported by Otto Stengel (1826), who documented the course of the disease in two sisters and two brothers of apparently healthy parents: the first symptom was loss of vision at an early age, followed by deterioration in cognition and speech, seizures and premature death.

The course of JNCL

JNCL is characterized by a severe loss of vision which becomes noticeable around age 4–8 years, with a developmental course that includes blindness, epilepsy, speech and language problems, cognitive regression and motor coordination problems. Individuals with JNCL may also experience a number of non-defining symptoms, such as problems with sleep, eating, breathing, pain, and mood (Rokne, 2009). JNCL is a complex disease and the many symptoms and declines entail severe challenges for the individuals with this disease and their families.

JNCL influences all aspects of development but it is vision problems together with dementia that characterize the disease. Visual impairment is usually the first sign and to a large extent contributes to the identification of the disease. However, most individuals with congenital or early blindness have developmental functioning that is comparable to their peers with normal sight. Unlike those groups, children with JNCL experience the onset of dementia not long after the onset of visual impairment. The childhood dementia, meaning the onset is in childhood rather than later in life, has a pervasive influence on development and learning, and compounds the difficulties imposed by the visual impairment. Childhood dementia is not well researched but knowledge from dementia research in general may be useful when planning educational and other interventions for this group. The consequences of blindness and dementia are therefore *the leading theme* that recurs throughout the book.

Figure 1.1 shows the average age at first occurrence of symptoms and difficulties typical of JNCL, based on information from parents whose children had received a JNCL diagnosis and participated in the present project (Appendix A).

There was considerable variation in age at onset, with reports of some symptoms and difficulties occurring mainly within a limited age span, and others exhibiting more variation. Symptoms falling within a more limited age span included the appearance of something being wrong with the child between the age of five and seven years, visual impairment became noticeable between five and eight years, difficulties following mainstream education between six and ten years of age, and memory problems when the child was between seven and twelve years old. The symptoms and problems showing more variation in when they first emerged included problems with speech, gait and balance, which were first observed when the participants were between 10 and 18 years of age, and motor problems with hands and arms, which tended to occur between the age of 14 and 21 years (see Chapter 7).

The onset of some problems and declines occurred in a rather fixed order while the sequence of other symptoms varied more from participant to participant. For instance, visual impairment and memory problems were early symptoms of the

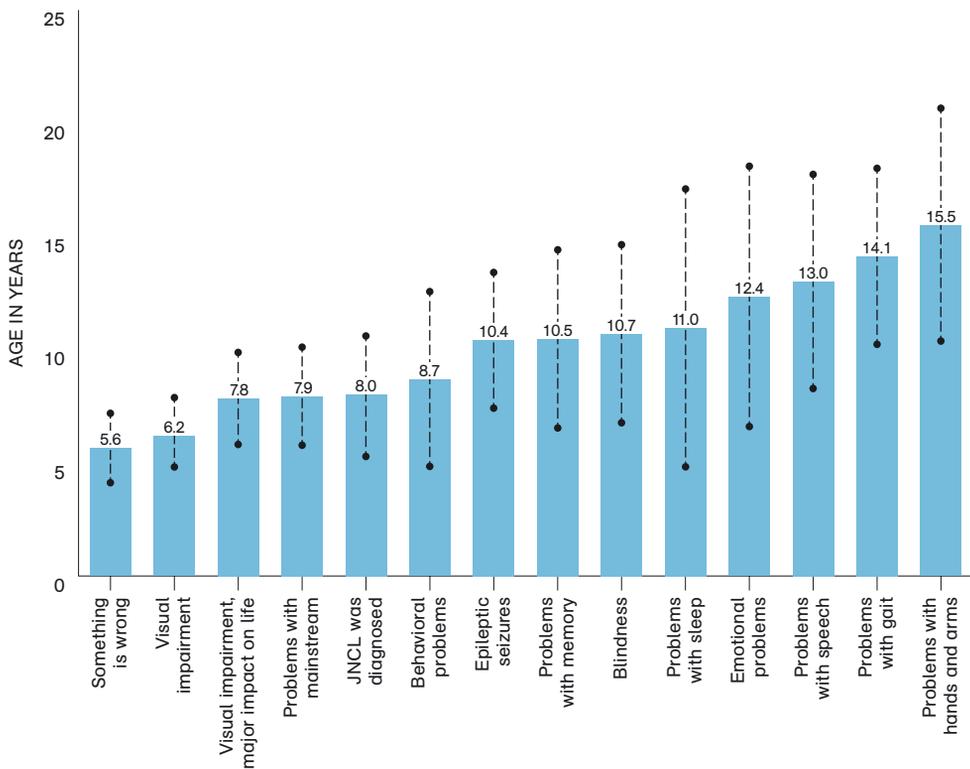


Figure 1.1 Average age at onset of some problems and declines
Note: The spotted lines represent standard deviations.

disease; the visual problems usually appeared earlier than the memory problems, while speech and motor problems were observed later. In some participants, problems with spoken language occurred before motor difficulties, while others showed the opposite sequence. However, there was also variation within the motor domain, most participants showed problems with gait and balance before problems with hands and arms.

All symptoms had not been observed in all participants. Figure 1.2 shows the percentage of participants who had shown the symptoms and difficulties related to the disease. Problems with speech, memory and gait and with following mainstream education had been observed in almost all participants. It may be noted that some symptoms appeared relatively late and a lower prevalence may be due to the age distribution in the sample. For example, about half of the participants had problems with fine motor skills (use of hands and arms) but these problems tended to appear rather late, on average around 15 years (see Chapter 7) and many participants had not reached this age yet. On the other hand, behavioral problems were first observed at an average age of eight years but comprehensive behavior problems were reported in only about half of the participants (see Chapter 27).

In addition to the common problems related to JNCL, parents had also observed a number of less frequent problems. Some parents with older children reported that their child had developed hypersensitivity for certain sounds or

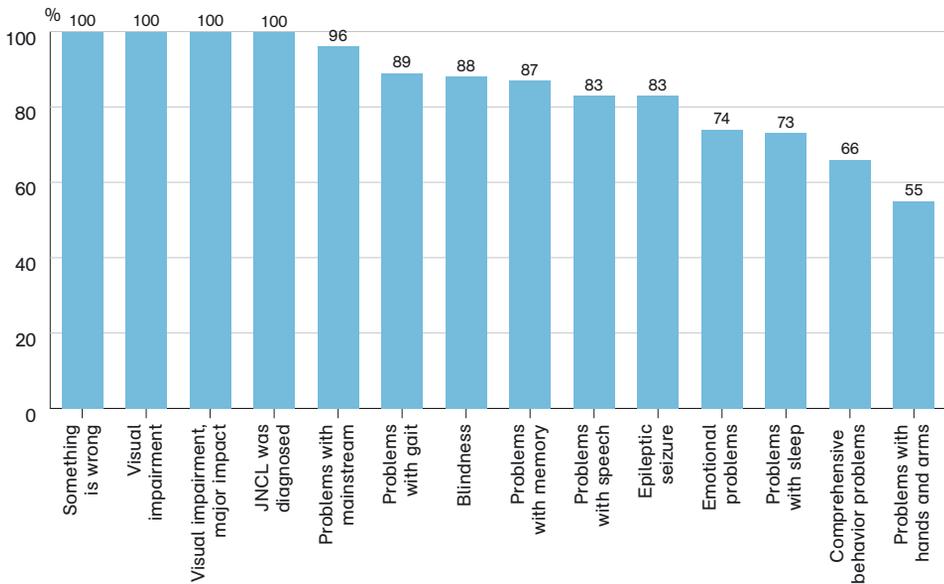


Figure 1.2 Prevalence of some observations, problems and declines (percent of total sample)
Note: Prevalences will be influenced by the age distribution in the sample.

an inability to cope with certain sound frequencies and noisy environments, or reacted strongly to non-anticipated sounds. Other parents and educational staff expressed that the participants' behavioral and emotional reactions were their biggest daily challenges (see Chapter 27).

Evidence-based practice

There is a general demand that practice should be evidence-based, and that assessment and intervention should have documented effects. Evidence-based practice means that there is a research basis for the assumption that the particular methods used in educational and other interventions for a child or adult are effective and useful (Berninger, 2015; Kratochwill & Shernoff, 2004; Stoiber et al., 2016). Studies where similar individuals are randomly allocated to a group that gets a new intervention or to a non-intervention or intervention-as-usual group, are considered the highest level of evidence. This kind of study is important for comparing different intervention methods or educational strategies. However, it is not always possible to implement such studies with children who have rare diseases. Individual case studies and systematic collection of professionals' experiences with the intervention methods used in practice may therefore be important for developing guidelines for best practice with these children and adolescents. Case studies can go into greater depth and provide insight into the processes underlying the atypical development and the intervention, also in a scientific sense (see Parker & Hagan-Burke, 2007; Yin, 2009). This requires that there is documentation and analysis of current practice, both of which are lacking for many groups of children with rare conditions.

The lack of documentation and systematic evaluation of experiences with different educational strategies and other interventions was an important motivation for the present project on JNCL and education. To our knowledge, systematic collection of experiences from the education of students with JNCL is lacking. Gathering information from professionals who are responsible for the education of a particular group, and from parents, represents a first step toward evidence-based practice. The results presented in this book may not meet the scientific requirements for strong evidence but they represent a first step toward systematization of the experiences of a relatively large group and may inspire further research in this area.

Raising the question whether there exist supportive research results for a particular practice is an opportunity to reflect on the methods used and hence to renew or supplement these methods. Some interventions continue to be useful, others fail the "evidence test" and may be replaced with approaches that

have a better theoretical and empirical basis. New knowledge about children's development and disorders may open up for new interventions but good practice is not only a matter of evidence but of professionals having an investigative and critical stance and reflecting on their own practice (Kratochwill, 2012). One aim of the present book is to encourage this kind of reflection on the approaches used in education and intervention for children, adolescents and adults with JNCL.

Education as a leading activity from childhood to emergent adulthood

Learning is a core element in adaptation. Learning may be defined as an experienced-based process which leads to relatively permanent changes in the knowledge, skills and behavior of an organism, and which is not caused by maturation, disease, fatigue or injury (Kolb, 1984). The transfer of knowledge across generations is basic to the evolution of human societies, and the school is society's main organization for providing shared knowledge and competence to all individuals, as well as ensuring the diverse competence society needs (see Chapters 9 and 12). However, humans also learn from their actions and exploration outside school. Much of this learning is social, as more competent members of the society provide guidance toward what is important and relevant in their society (Tomasello, 2009; Vygotsky, 1978). The individual learns throughout the lifespan but the acquisition of new knowledge and skills is especially important in childhood and adolescence. In new situations, adults can to a greater extent use knowledge and skills they already have.

Abilities and learning vary between members of a society and some have problems that imply an atypical course of development (von Tetzchner, 2019). Both the content and method of teaching and the students' ways of learning will vary with the physical and mental abilities and skills of the students. In spite of such differences, educational activities are equally important to students with typical and atypical development, including to students who show regression in abilities and for whom maintenance of knowledge and skills, rather than new knowledge and skills, becomes the major aim.

JNCL and education

Over the last decade, there has been some attention to the processes involved in educating children and adolescents with JNCL, especially after the first international conference on JNCL and education (von Tetzchner, 2006). Some useful approaches appear to be available but there is a lack of evidence in relation

to the educational strategies that are currently in use, and there has been no comprehensive evaluation of the strategies that are applied. There are very few publications about JNCL with an educational perspective (von Tetzchner, Fosse & Elmerskog, 2013). Research publications about cognitive functions in children and young people with JNCL rarely mention any implications the findings may have for education. Medical, psychological and educational professionals seem to have focused on the disease itself and its consequences for the individual's development, but not on how it affects participation in education, including both challenges and possibilities. There is thus still limited knowledge about the typical learning paths and variation within this group, indicating a need for larger studies.

The complex picture of different symptoms and problems apparent in the present study (see Figure 1.1.) indicates a need for competence in different areas. Some symptoms, declines and problems were affecting all participants, whereas others were affecting only some of them. The declines are caused mainly by neurological disorders but some problems are related to environmental factors. The two main roles of education and learning are to compensate for declines caused by the neurological progressions, and to reduce environmental influences that may cause secondary problems. The challenges are many and complex, but most challenges can be met by educational measures for maintaining skills and sustaining participation and quality of life for the individual. The main challenge for educators is to provide the right interventions at the right time based on knowledge and experience.

A major focus in this book is the role of dementia in the development of individuals with JNCL and its consequences for education. Awareness of childhood dementia in education highlights the problems related to observing and measuring the effects of interventions. A person with dementia will – independent of age – always show declines and loss of functions even with the best educational interventions. Education has standardized tools and observations for measuring learning effects in students who show progress, but few or no standard instruments for observing the effects of educational interventions for low-frequency groups like students with JNCL, who are showing declines.

Educational services for individuals with JNCL are in most countries organized under visual impairment because visual impairment is a basis for the diagnosis (see Chapters 3 and 9). This situation is also reflected in the organization of the present project. Most of the project members are affiliated with institutions in the field of visual impairment. This implies that the loss of vision is usually met with the necessary competence, even if many mainstream schools have limited competence and are struggling in this area. Declines in other areas of the child's functioning (e.g., speech and language, cognition, and motor performance) are often not met with the expertise needed. Services for children and young people with JNCL need

to be supplemented by interventions that are not usually part of interventions related to visual impairment. Education and other interventions for students with JNCL should also be based on an understanding of how different declines and problems interact in forming the developmental course, and of possible measures and strategies that can meet these combined challenges. For instance, emotional and behavioral reactions may be related to the unavoidable visual and cognitive decline, but also to a lack of cautionary and compensatory measures that might have contributed to making the changes less stressful and upsetting.

There is little documentation of the role of education and learning for children and adolescents with JNCL and a need for a larger body of histories about students with JNCL with typical as well as exceptional educational paths. The brief case histories presented in this book illustrate that education may have a significant impact on the life situation for individuals with JNCL. Asking what factors were significant for the long-term educational success of these individuals is a valuable start, but it should be emphasized that those factors mentioned by informants may not be representative for students with JNCL in general. However, they do demonstrate possibilities for individuals in this group and the importance of providing person-centered services (see Chapters 11 and 12). One common factor in successful case histories seems to be that the persons with JNCL established strong interests early in life, interests that could be used in education to promote the development of knowledge and skills. The students' education was characterized by continuity, with each school knowing and building on the work of the former school. There was close collaboration between families, schools and multidisciplinary teams and the resources that were provided were necessary for continuity in the long-term and goal-oriented education. The important role of the parents in the present case histories cannot be overestimated; they were the experts on their own children and the final guarantors for life flow and continuity. Case histories show the importance of starting adult life with a rucksack and an agenda full of activities, interests and skills. Personal style and image must be respected by the people who take over the responsibilities for their adult life. This means that their sheltered workplace and new home should take each individual's strengths, weaknesses and life history into account.

As the present book demonstrates, there is much to learn about the education of children and adolescents with JNCL, and its possible consequences for adult life. Part of the knowledge comes from the observed variation in perceptual, motor, cognitive and language abilities in this and some other diseases with cognitive decline in childhood (Schoenberg & Scott, 2011). However, the experiences of families with members who have JNCL and the professionals who support the children's learning and coping, are important sources of information. Information from parents and educational staff is the empirical foundation of the present

project (Appendix A). Research on education usually focuses on teaching and learning during the period of formal education, as defined in each country, but there is a need to include some consideration of the long-term consequences of the education and of the intervention strategies and adaptations that may contribute to positive adulthood. Some studies include emotional and behavioral reactions to educational and other aspects of daily life (Bills, 2011; Bäckman, Santavouri, Åberg & Aronen, 2005). Adaptations provided to the educational settings of people with JNCL must facilitate learning possibilities while minimizing negative challenges. Only then will they contribute significantly to improved learning conditions, better maintenance of skills, less frustration, and hence less severe behavioral and emotional reactions in children as well as adults (see Chapter 27).

Registry

Due to the low prevalence of JNCL, many teachers and other professionals know only one or two students with this disease. In order to avoid self-fulfilling stereotypes, there is a need to gain information about the variation in the course of the disease and how teachers and professionals can evaluate developmental signs and prepare for the potential progression and regression in the individual student's developmental course (see also Chapters 2 and 10). Today, there are registries for many diseases and disabling conditions, such as for Duchenne muscular atrophy (www.duchenneregistry.org). The data collected here might be a first step toward developing such a registry for JNCL and other CLN diseases. It would be a natural task for national or regional competence centers for JNCL to register and systematize educational strategies and teacher experiences within their geographical area. International collaborative research efforts may compare the development of students with JNCL in different educational (e.g., mainstream and special schools) and cultural settings (see Chapter 9). Professionals and families can collaborate with universities to ensure appropriate theoretical and methodological competence.

Overview

The chapters in this book describe the features of JNCL with an emphasis on vision impairment and early onset dementia, and discuss a range of theoretical, empirical and practical issues related to educational and other supports for children, adolescents and adults with JNCL. Chapter 2 first relates the developmental course of JNCL to developmental theory in general, as well as to Baltes' theory of goal

selection, optimization and compensation, and second, it introduces the concept «zone of developmental maintenance» as a tool for understanding the variation in development among individuals with JNCL and supporting intervention planning. Chapter 3 provides an introduction to medical issues related to JNCL. The chapter is rather short because there is a huge literature on the biological and physiochemical aspects of the disease and the main aim of this book is to provide information about educational possibilities. Chapters 4–7 describe the four main domains related to JNCL: development and decline in vision, cognition, language and motor abilities. Chapter 8 discusses ethical issues related to working with people with disabilities and developmental decline. Chapters 9–12 are about the basic processes related to assessment, planning, organization and implementation of educational and other non-medical interventions for individuals with JNCL, with special attention to proactive, precautionary and hastened teaching and learning. Chapters 13–21 present more specific teaching areas and educational approaches, especially related to reading, writing and mathematics, but also include the use of technology, games, drama and music. An important consideration addressed in these chapters is the need for support personnel to recognize how for each child with JNCL there is a process of moving from independent to interdependent functioning while participating in educational activities and coping in everyday life. Chapter 22 discusses the consequences of JNCL for peer interaction, social life and participation in society. Chapter 23 goes beyond the school years and discusses processes related to the transition from a school life to an adult life. Chapter 24 addresses parent needs and support, and chapter 25 presents experiences of being a brother or sister of a person with JNCL. Chapter 26 describes the important functions of the family associations. Finally, Chapter 27 discusses the behavioral and emotional reactions that may be observed in individuals with JNCL; the relationship between these reactions to the developmental course of the disease and the complexities of the difficult life situations that ensue; and strategies that may lessen the stress and frustration of the persons, and thereby their behavioral and emotional reactions.

References

- Åberg, L., Lauronen, L., Hämäläinen, J., Mole, S. E., & Autti, T. (2009). A 30-year follow-up of a neuronal ceroid lipofuscinosis patient with mutations in CLN3 and protracted disease course. *Pediatric neurology*, *40*, 134–137.
- Bäckman, M. L., Santavuori, P. R., Åberg, L. E., & Aronen, E. T. (2005). Psychiatric symptoms of children and adolescents with juvenile neuronal ceroid lipofuscinosis. *Journal of Intellectual Disability Research*, *49*, 25–32.

- Baltes, P. B., & Baltes, M. M. (1990). Psychological perspectives on successful aging: The model of selective optimization with compensation. In P. B. Baltes & M. M. Baltes (Eds.), *Successful aging: Perspectives from the behavioral sciences* (pp. 1–34). Cambridge: Cambridge University Press.
- Bennett, M. J., & Rakheja, D. (2013). The neuronal ceroid–lipofuscinoses. *Developmental Disabilities Research Reviews*, 17, 254–259.
- Berninger, V. W. (2015). *Interdisciplinary frameworks for schools: Best professional practices for serving the needs of all students*. Washington, DC: American Psychological Association.
- Bills, W. (2011). *Teach and be taught*. Reynoldburg, OH: Batten Disease Support and Research Association, 2011. Retrieved June 1st 2012 from <http://www.bdsra.org/programsservices/bdsra-publications/>
- Cooper J. D. (2010). The neuronal ceroid lipofuscinoses: The same, but different? *Biochemical Society Transactions*, 38, 1448–1452.
- Haltia, M., & Goebel, H. H. (2013). The neuronal ceroid-lipofuscinoses: A historical introduction. *Biochimica et Biophysica Acta (BBA)-Molecular Basis of Disease*, 1832, 1795–1800.
- Kohlschütter, A., & Schulz, A. (2009). Towards understanding the neuronal ceroid lipofuscinoses. *Brain and Development*, 31, 499–502.
- Kousi, M., Lehesjoki, A. E., & Mole, S. E. (2012). Update of the mutation spectrum and clinical correlations of over 360 mutations in eight genes that underlie the neuronal ceroid lipofuscinoses. *Human Mutation*, 33, 42–63.
- Kratochwill, T. R. (2012). Comments on «Distinguishing science from pseudoscience in school psychology»: Evidence-based interventions for grandiose bragging. *Journal of School Psychology*, 50, 37–42.
- Kratochwill, T. R., & Shernoff, E. S. (2004). Evidence-based practice: Promoting evidence-based interventions in school psychology. *School Psychology Review*, 33, 34–48.
- Mole, S. E., & Haltia, M. (2015). The neuronal ceroid-lipofuscinoses (Batten disease). In R. N. Rosenberg & J. M. Pascual (Eds.), *Rosenberg’s molecular and genetic basis of neurological and psychiatric disease, Fifth edition* (pp. 793–808). New York: Academic Press.
- Mole, S. E., Williams, R. E., & Goebel, H. H. (Eds.) (2011). *The neuronal ceroid lipofuscinoses (Batten disease)*. Oxford, UK: Oxford University Press.
- Parker, R. I., & Hagan-Burke, S. (2007). Useful effect size interpretations for single case research. *Behavior Therapy*, 38, 95–105.
- Rakheja, D., & Bennett, M. J. (2018). Neuronal ceroid-lipofuscinoses. *Translational Science of Rare Diseases*, 3, 83–95.
- Rider, J. A., & Rider, D. L. (1999). Thirty years of Batten disease research: Present status and future goals. *Molecular Genetics and Metabolism*, 66, 231–233.
- Rokne, S. (2009). *Hver dag teller. En orientering om Spielemeyer-Vogs sykdom* (Every day counts. An introduction to Spielmeyer-Vogt disease). Asker, Norway: Norsk Spielmeyer-Vogt Forening.
- Schoenberg, M. R., & Scott, J. G. (2011). Cognitive decline in childhood or young adulthood. In M. R. Schoenberg & J. G. Scott (Eds.) *The little black book of neuropsychology: A syndrome-based approach* (pp. 839–861). New York, NY: Springer.
- Stengel, C. (1826). Beretning om et mærkeligt Sygdomstilfælde hos fire Sødskende i Nærheden af Røraas (*The history of a strange case of disease in four siblings near Røros*). *Eyr et Medicinsk Tidsskrift*, 1, 347–352.

- Stoiber, K. C., Purdy, S., & Klingbeil, D. A. (2016). Evidence-based practices. In N. N. Singh (Ed.), *Handbook of evidence-based practices in intellectual and developmental disabilities* (pp. 41–68). Cham, Switzerland: Springer.
- Tomasello, M. (2009). *Why we cooperate*. London, UK: MIT Press.
- von Tetzchner, S. (Ed.) (2006). Proceedings of the First International Education Conference on Batten Disease. Örebro, Sweden: Swedish Institute for Special Needs Education.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- von Tetzchner, S., Fosse, P., & Elmerskog, B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta (BBA)-Molecular Basis of Disease*, 1832, 1894–1905.
- Vygotsky, L. S. (1978). *Mind in society: The development of higher mental processes*. Cambridge, MA: Harvard University Press.
- Williams, R. E., & Mole, S. E. (2012). New nomenclature and classification scheme for the neuronal ceroid lipofuscinoses. *Neurology*, 79, 183–191.
- Yin, R. K. (2009). *Case study research: Design and methods, Fourth edition*. Thousand Oaks: Sage.

2

A Developmental Perspective on Juvenile Neuronal Ceroid Lipofuscinosis

Stephen von Tetzchner

Juvenile neuronal ceroid lipofuscinosis (JNCL) is a developmental disorder, with a gradual loss of vision and other functions, beginning in childhood. The pathological biological bases of JNCL (see Chapter 3) constrain developmental growth and eventually lead to developmental decline in most areas. The present chapter presents a theoretical approach to framing the developmental processes involved in the regression, arguing that a developmental way of thinking is useful even when there is developmental decline. This chapter examines how a person with increasing cognitive impairment grows and develops, by addressing multiple aspects and placing them in different frameworks of understanding. The theoretical model and knowledge provided in this book, about the typical course and developmental variation in JNCL, provide a basis for timely and appropriate education and other forms of interventions. This approach may contribute to compensating for functional loss and decline in individuals with JNCL through education, adaptation and support within their «zone of developmental maintenance». This approach may be adequate for children with other childhood dementia disorders.

The concept of development

Traditionally, development has been defined as an age-related process involving positive changes in the structure and functioning of human beings and animals as a result of interaction between biological structures, psychological states and ecological factors. At the core of this process lies *transformation*: something new emerges, less may become more, simplicity may turn into complexity, and limited skills may evolve into advanced mastery (Overton, 2015). Development towards adulthood implies a greater degree of autonomy and independence from the parents and a successively increasing social affiliation in the society (Keller,

2016). However, from a life-span perspective, developmental transformation also includes reductions of skills and abilities (Baltes & Baltes, 1990).

Development is thus the conglomeration of processes that together lead to an individual's physical, cognitive and personal characteristics, social relationships and roles in society. Change is a defining characteristic of development but in most areas there is both change and continuity. The individual's mastery and understanding of the world change, but change always emerges from a foundation of prior abilities and experiences, and the individual remains the same (Nelson, 2007). The parallel paths of change and continuity are also a characteristic of developmental decline, even if the process of regression differs from *typical development* in many aspects.

There is considerable individual variation in most aspects of development. Typical development is the most common course, with unimpaired functions and ordinary individual differences between children. *Atypical development* is a broad term used to describe all forms of irregular development, such as the normal but unusual language development of deaf children when they learn sign language instead of spoken language, or the different ways of thinking and reasoning that differentiate children with autism spectrum disorder from their peers. Development follows the same basic principles, regardless of whether it proceeds typically or atypically, but while the organism adapts to its environment, the environment must also have properties that allow the organism – with its strengths and weaknesses – to develop. Most children with typical development will show positive development in quite different environments. Children with atypical development have a narrower range of possibilities and are more dependent on an environment that fits their particular strengths and weaknesses (von Tetzchner, 2019).

Development and disability

Some children acquire skills and abilities early on, while others have late or unusual development in one or more areas. Some have impairments or deficits that may inhibit some or all aspects of development, rendering them unable to perform many actions or requiring them to perform actions in unconventional ways.

In the gap model (Lie, 1996), disability is defined as a gap between an individual's abilities and the abilities that the environment requires to allow participation. Both the individual's abilities and the demands of society change with the individual's age and training, and hence the disability gap may both increase and decrease over time. The gap in functioning between individuals with severe disabilities and those with typical functioning may widen with age (Stadskleiv, 2017). However,

the developmental consequences are not determined by an impairment or deficit alone, but instead, both primary and secondary consequences are related to the support provided by the family, professionals, and society at large. There are aspects of disability that reflect structure and process. The *structural* perspective is a characterization of the child's actual skills and abilities – or his or her lack of skills and abilities. The *processual* perspective characterizes processes related to both a primary impairment or deficit, and the possible secondary consequences of these. From a developmental point of view, disabilities emerge progressively or regressively as a result of a comprehensive and complex process, where learning is an adaptation to the ecology, culture and life situation of the individual. There is no simple relationship between an impairment and its developmental consequences. Intervention may lose efficiency if designed on the basis of the specific impairments of the child, out of context; rather, design should proceed according to both the child's functioning and his or her physical and social environment. For example, when a child is unable to perform self-propelled movement, it has consequences for the child's exploration, world knowledge, concept development, self-regulation and attachment (Anderson et al., 2013; Campos et al., 2000). Similarly, the memory impairment accompanying dementia not only affects thinking and reasoning, but also many other skills including communication, opportunities for learning and socialization, and maintenance of other skills and abilities.

Development is transactional

Children are dependent on adults to support their development of abilities and autonomy, and parents usually form the core of children's social environment. However, it is not only the case that the environment influences the child's development, but also that the environment is influenced by the child. For example, extroverted and social children elicit very different reactions from the people in the environment than introverted and shy children (Kagan & Snidman, 2004). Children of small stature are met differently than children who are large for their age (von Tetzchner, 1998). Children who are blind may be listening actively but without facing their parents, and may therefore be perceived as inattentive and uninterested by the parents (Fraiberg, 1977).

Transaction designates the reciprocal interaction between the child and the environment (Sameroff, 2010). The environment influences the child, and the child, in turn, influences the environment. The changed environment without facing the parents influences the child, who in turn influences the environment, and so on. This means that the child's development is influenced both by how the child perceives the environment *and* by how the child is perceived by the environment. People in the environment will be influenced by the child and

adapt to the child's characteristics as they perceive them. Transaction thus has a subjective basis. Transaction is a basic feature of development, whether it involves motor skills, personality, social relationships, emotions, communication, problem solving or other areas.

Parenting styles reflect parents' experience, beliefs about children and childhood, and sensitivity and ability to adapt to their child's needs and abilities. As the child develops, the parents gain detailed knowledge about the general and unique characteristics of their child, the child's possibilities and limitations. However, parents' experience and general beliefs do not usually include children with atypical development, and they may lack knowledge about how to interpret and react to a child who develops differently than most children. Their expectations and interpretations of their child with a disability may therefore be guided by their general knowledge about children, together with the knowledge they have about their own child and the child's disability. Parenting may still be positive for the child, but parenting of children with atypical development often reflects both overestimation and underestimation, because the parents do not have sufficient knowledge about the typical course and variation that the impairment of their child may imply. As the child grows, parents of children with disabilities also get a better understanding of the child's possibilities and limitations. However, it can still be difficult for parents to understand both what their child with a disability can do, as well as what the child cannot do. When parents lack sufficient insight into the child's development, abilities and challenges, their expectations may not match the possibilities and limitations of the child, and they may misinterpret the child's behavior and reactions. They may need to be guided by professionals towards a *redefinition* of the child's behavior and expressions, of his or her biological possibilities and impediments and of how they can create an environment that adapts to both the abilities and disabilities of the child throughout development (Sameroff & Fiese, 2000). Mobility supports the child's exploration, world knowledge, concept development, self-regulation and attachment independent of the manner in which the child moves around (von Tetzchner, 2019). Also teachers and other professionals may need guidance to understand and become able to provide appropriate developmental support to a child with a disability. In particular when the child has a rare disorder, many professionals will have no prior experiences with that disorder, and in addition may spend only a few hours together with the child every week. They may need considerable guidance and support to become able to create a physical, educational and social environment that promotes an optimal development.

Development may imply both progression and regression

Abilities and disabilities do not come ready-made; instead they are the result of complex interactions between biological and environmental processes over time. Development may include periods with reduction in skills and abilities, and intervention must be based on an understanding of the process. The developmental changes in regression are age-related but the direction is different from developmental progression and reflect complex processes. In particular, the intervention measures needed to support adaptation to weaker physical, perceptual and cognitive abilities in children and young people may be poorly known and understood. It may be difficult to define future goals and make decisions about intervention when regression is observed or expected, and to evaluate the effects of the interventions when the functional level of the individual has become lower.

Selection, optimization and compensation

Growing up with a disability represents a way of life that in important ways differs from that of most children in the community. Children with JNCL, as compared with their peer group, typically have to find other ways to play and select alternative leisure activities. Some children have few choices for both themselves and the people who are making adaptations to their environment. The differences between the developmental trajectories of children with JNCL and other children, with or without disabilities, are increasing with age.

In the lifespan theory of Paul and Margret Baltes (1990) human development is described as processes of selection, optimization and compensation (SOC). These theoretical concepts are particularly useful for explaining development in individuals who are lacking or losing many core abilities. *Selection* concerns the individual's choice of relevant and achievable goals, while *optimization* is the process of forming and maintaining the means to achieve the goals that have been selected implicitly or explicitly. *Compensation* is the use of new strategies to maintain functions and goals that are no longer sustainable in ordinary ways, for example due to physical weakness and illness, which may prevent or delay some of the functional decline (Baltes, 1987, 1997). The model has been applied mainly to developmental processes in elderly adults, but it includes development throughout the lifespan, including early development and development in individuals with negatively progressing conditions (Baltes & Smith, 2004).

The concepts of selection, optimization and compensation represent a useful theoretical framework in the present context because they highlight important aspects of the developmental process in individuals with declining functioning

in core areas. For example, a child or adolescent with JNCL may together with parents and professionals select actions and activities that can scaffold «learning in participation». Optimization may include guidance of adults and peers in the environment, and compensation may include special training as well as adaptation of the classroom and educational strategies. From a developmental perspective, the aim of early intervention is not primarily to «repair» functions but to initiate and give direction and support to the developmental processes, preventing decline and supporting coping maintenance. For example, when a child is unable to perform self-propelled movement, a wheel chair provides access not only to movement itself, but also movement that results in participation opportunities.

The zones of proximal development and developmental maintenance

Children gradually learn to master new things. Many of the things they are unable to do independently, become possible in collaboration with adults and more competent peers. The things children are unable to master on their own but are able to accomplish in collaboration with others are within what Vygotsky (1962) calls their zone of proximal development. The beginning and end of the zone of proximal development are defined by what a child is able to achieve, respectively independently and with help. The point at which children no longer master a task on their own marks the beginning of the zone. When a maximum amount of help from more competent peers and adults does not enable the child to attempt to solve a task, the task lies outside the child's zone of proximal development.

It is within the zone of proximal development that learning can take place, and adults must be able to introduce new «tasks» and adapt their mediation to the child's zone. If a child encounters a task with a degree of difficulty that is beyond this zone, the child will not understand the task and thus will not be able to contribute to its solution. If the task is too easy, solving it will not contribute to the child's development or changes in their zone of proximal development. For tasks located within the zone, the strategies contributed by adults are termed *scaffolding* (Wood, Bruner & Ross, 1976). Scaffolding corresponds to the principle of *guided participation*, that is, that children develop while participating actively in culturally valued activities with the guidance, support and challenge of children and adults who vary in skill and status (Hundeide, 2003; Rogoff, 1990).

Scaffolding is mediation: adults attempt to clarify what is required to solve a task, limit those aspects of the task that the child is unable to master – for example by holding an object stationary while the child tries to take something from it – and shield the child from distractions that can draw attention from the task at

hand. As the child develops, the help and support are gradually reduced, and once the child is able to master a task independently, scaffolding is no longer needed. This means that adults guide the child towards tasks at the child's own level *as well as* offer help that the child actually benefits from. Too little help can prevent children from being able to solve the task. Too much help can reduce children's trust in their own ability, because the help provided by adults also reflects their evaluation of the child's competence (von Tetzchner, 2009). Both the task and help must lie within a child's zone of proximal development if adults are to contribute to the child's development. The transactional influences are working through the adults' sensitivity to the child and their ability to create an environment that the child can cope with and learn from. Children with disabilities have fewer social opportunities and more constraints in everyday life than most children, but their development is still facilitated by participation in social activities.

The concept of zone of proximal development applies to developmental progression, and the aim of scaffolding is to promote the child's future independent mastery. This aim cannot be applied to developmental regression. For an individual who is showing developmental decline, guidance and support therefore have a different function than for children showing progression. The aim of adults' help is to contribute to maintaining the individual's ability to manage tasks that he or she can no longer master independently. Because scaffolding may support prolonged self-initiated performance and autonomy, it is within the individual's *zone of developmental maintenance* (see Figure 11.1, page 193). When the action is performed only by the adult without any contribution from the individual with developmental decline, the task is outside the zone of maintenance. Like in the zone of proximal development, maintenance through interdependence (see Chapter 16) depends on the knowledge and sensitivity of the adult in recognizing and providing the individual with the help that he or she needs, not too much and not too little.

Scaffolding reflects the transactional nature of development, being characterized by the adult being sensitive and giving help and support that are adapted to the skills and abilities of the individual. For children and young people with developmental decline, the scaffolding strategies applied by parents and professionals will reflect how they perceive the abilities and limitations of their child and the adaptive nature of ordinary or compensatory strategies used to maintain or support a particular function. An individual with JNCL may need help solving immediate challenges, but the main aim of scaffolding of individuals with JNCL is to delay the decline and maintain their functions and coping as long as possible, by increasing the scaffolding when needed. According to social constructivist developmental theories, adults enable children to acquire writing, arithmetic, and other cultural cognitive tools. For individuals with JNCL, the

cognitive tools developed early in life may play an important strategic role in maintaining functions and delaying later decline (see Chapter 14).

Scaffolding is often perceived as a «natural» rather than educational process, emerging from the interactions between children and their caregivers. For individuals with JNCL, intervention may also represent a scaffolding activity, for example when it promotes communicative success in social interactions that are not easily guided in ordinary situations. Moreover, for children with JNCL, the developmental process will have to be more planned and the scaffolding more explicit and direct than for children without such a comprehensive disability. Scaffolding in the zone of maintenance is integrated with the processes of selection, optimization and compensation.

The role of planning in atypical development

A common feature of atypical development is the need for planning. Although nearly all children attend schools with planned education, children with impairments are more dependent on detailed plans than children with typical development (see Chapter 11). The choices made by the family are closely interwoven with decisions taken by professionals, and often made through dialogue (see Chapter 24). The environment must be arranged individually and plans may be necessary for creating opportunities and reduce the restrictions on participation in activities that typically characterize the environment of individuals with JNCL (see Chapter 16).

Good planning requires sufficient knowledge of the developmental processes and the consequences impairments may have in the short and long term. Integrating the theory of Baltes and scaffolding, one important task of professionals is to guide individuals with JNCL and their families in selecting and optimizing goals. All children and adolescents thrive with goals that challenge, as long as they are comprehensible and achievable. However, for children and young people who show decline, goals are often *not* developed on the basis of a thorough assessment or systematic observations of the child's current abilities, up-to-date knowledge about the condition and professional reflection. The result may be a lack of appropriate compensating measures and goals. The goals may be either too repetitive and boring or too new and complex, sometimes leading to passivity or challenging behavior in the child. Plans and goals should include precautionary measures and be based on an anticipation of how JNCL is likely to affect the development and learning of the child in the future.

References

- Anderson, D. I., Campos, J. J., Witherington, D. C., Dahl, A., Rivera, M., He, M., et al. (2013). The role of locomotion in psychological development. *Frontiers in Psychology*, 4, 440.
- Baltes, P. B. (1987). Theoretical propositions of life-span developmental psychology: On the dynamics between growth and decline. *Developmental Psychology*, 23, 611–626.
- Baltes, P. B. (1997). On the incomplete architecture of human ontogeny: Selection, optimization, and compensation as foundation of developmental theory. *American Psychologist*, 52, 366–380.
- Baltes, P. B., & Baltes, M. M. (1990). Psychological perspectives on successful aging: The model of selective optimization with compensation. In P. B. Baltes & M. M. Baltes (Eds.), *Successful aging: Perspectives from the behavioral sciences* (pp. 1–34). Cambridge: Cambridge University Press.
- Baltes, P. B., & Smith, J. (2004). Lifespan psychology: From developmental contextualism to developmental biocultural co-constructivism. *Research in Human Development*, 1, 123–144.
- Campos, J. J., Anderson, D. I., Barbu-Roth, M. A., Hubbard, E. M., Hertenstein, M. J., & Witherington, D. (2000). Travel broadens the mind. *Infancy*, 1, 149–219.
- Fraiberg, S. (1977). *Insights from the blind*. New York: Basic Books.
- Hundeide, K. (2003) *Barns livsverden: Sosiokulturelle rammer for barns utvikling, Andre utgave* (Children's lifeworld: Sociocultural frames for child development, Second edition). Oslo: Cappelen Akademisk.
- Kagan, J., & Snidman, N. C. (2004). *The long shadow of temperament*. Cambridge, MA: Belknap Press.
- Keller, H. (2016). Psychological autonomy and hierarchical relatedness as organizers of developmental pathways. *Philosophical Transactions of the Royal Society B: Biological Sciences*, 371 (1686), 1–9.
- Lie, I. (1996). *Rehabilitering og habilitering: Prinsipper og praktisk organisering, Annen utgave* (Rehabilitation and habilitation: Principles and practical organization, Second edition). Oslo: Ad Notam Gyldendal.
- Nelson, K. (2007). *Young minds in social worlds: Experience, meaning and memory*. Cambridge, MA: Harvard University Press.
- Overton, W. F. (2015). Processes, relations, and relational-developmental-systems. In R. M. Lerner, W. F. Overton & P. C. M. Molenaar (Eds.), *Handbook of child psychology and developmental science, Seventh edition, Volume 1: Theory and method* (pp. 9–62). Hoboken, NJ: Wiley.
- Rogoff, B. (1990). *Apprenticeship in thinking – Cognitive development in social context*. Oxford, UK: Oxford University Press.
- Sameroff, A. J. (2010). A unified theory of development: A dialectic integration of nature and nurture. *Child Development*, 81, 6–22.
- Sameroff, A. J., & Fiese, B. H. (2000). Models of development and developmental risk. In C. H. Zeanah (Ed.), *Handbook of infant mental health, Second edition* (pp. 3–19). London, UK: Guildford.
- Stadskleiv, K. (2017). The relationship between language, expressive abilities and executive functioning in children with speech and motor impairment. Dissertation, University of Oslo, Norway.
- von Tetzchner, S. (1998). *Noonan syndrome. Characteristics, development, and intervention*. Oslo, Norway: Centre for Rare Disorders, The National Hospital.

- von Tetzchner, S. (2009). Suporte ao desenvolvimento da comunicação suplementar e alternativa (Scaffolding the development of augmentative and alternative communication). In D. Deliberato, M. de J. Gonçalves & E. C. de Macedo (Eds.), *Comunicação alternativa: Teoria, prática, tecnologias e pesquisa* (pp. 14–27). São Paulo: Memnon Edições Científicas.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- Wood, D., Bruner, J. S., & Ross, G. (1976). The role of tutoring in problem solving. *Journal of Child Psychology and Psychiatry*, 17, 89–100.

3

Medical Aspects of Juvenile Neuronal Ceroid Lipofuscinosis

*Alfried Kohlschütter, Jonathan D. Cooper,
Sara E. Mole, John R. Østergaard and Angela Schulz*

Juvenile neuronal ceroid lipofuscinosis (JNCL) is a brain disease with symptoms usually emerging slowly in late preschool age or in the early school years. The disease influences the brain, leads to epilepsy and deprives children of key abilities, such as vision, intellectual capacity, motor ability and language, described in other chapters of this book. Eventually, the disease leads to a decline in self-help skills and a shortened life span. The sequential loss of skills follows a characteristic order and contrasts with the typical development in all areas during puberty and adolescence. People who are caring for or involved in education of children and young people with JNCL need to be aware of the opposite processes of development and decline that are simultaneously at work in these young people (see Chapter 2). Understanding the medical basis of the disease and how it affects a child or young person's abilities will make it easier to react appropriately to the numerous challenges caused by the disease, both in everyday life and in education, which is the most important activity from late childhood to emergent adulthood. The impaired quality of life relating to JNCL varies considerably with the phase of the disease and the presence of other medical issues.

Definition of JNCL

Neuronal ceroid lipofuscinosis (NCL) is a group of different genetic disorders with many common features, both in respect to symptoms and underlying biological features. JNCL refers to an NCL disease that is «juvenile» and starts in early age. «Neuronal» means that neuronal cells (nerve cells) are mainly affected by the disease process and «ceroid lipofuscin» (Latin for «waxy, fatty and brownish») refers to a peculiar material that is found in large quantities in all cells and is characteristic of all NCL diseases.

Classic JNCL has been known for a long time, with the first description dating back to Stengel (1826). Other names for JNCL are Batten disease or Spielmeyer-Vogt disease, but using historical designations is not always adequate. For example, in parts of the Anglicized world, the term Batten disease is often used to designate the whole spectrum of NCL disorders, including forms that manifest in infants or in adults. Due to recent progress in genetic research, the medical classification of NCL disorders is currently based on the genes involved and the age of disease onset (Table 3.1). In this book, the term juvenile neuronal ceroid lipofuscinosis (JNCL) refers to all forms of NCL diseases with a juvenile onset.

Genetic causes of JNCL

All types of JNCL are caused by mutations (alterations) in various genes, creating different variants or alleles of the DNA material responsible for the hereditary transmission of personal characteristics. In the vast majority of children and young adults with JNCL, the mutated gene is called CLN3, and they may be said to be having juvenile CLN3 disease. There are other forms of NCL disease with a juvenile onset caused by mutations in CLN1 or CLN2, and very rarely in other genes (see Table 3.1).

There are other medical conditions with similarities to JNCL, such as the mucopolysaccharidoses (MPS disorders) or Niemann-Pick disease type C (NPC). These conditions differ from JNCL in the absence of visual loss and a more pronounced involvement of organs outside the nervous system (Schoenberg & Scott, 2011).

Inheritance of all JNCL forms is autosomal recessive, which means that the individual has received mutated genes from both their father and mother. As human beings carry a double set of most genes, a person with only one such NCL gene variant (a carrier or heterozygous person) does not usually develop symptoms of the disease because the second, non-mutated, gene compensates for the defective NCL gene. A child who has inherited a defective CLN variant of the NCL gene from both parents, either homozygous (when the mutations are the same) or compound heterozygous state (when the mutations are different), cannot compensate for the pathological function of the NCL gene, and therefore will develop the disease. One of the consequences of this form of inheritance is that a family can have several children who are affected by the disease. In the family described by Stengel in 1826, all four children developed the disease. A child will have a 50 percent probability of inheriting one abnormal gene, which would make the child a carrier who is unaffected by the disease. There is a 25 percent probability of the child being born with two non-mutated genes and hence

Table 3.1 Classification of currently known NCL diseases according to genes and age of manifestation

Disease Name according to mutated gene	Age at which first symptoms typically appear				
	At birth (congenital)	Before 1 year (infantile)	2–4 years (late infantile)	5–12 years (juvenile)	20–30 years (adult)
CLN1 Disease		X	X	X	X
CLN2 Disease			X	X	X
CLN3 Disease				X	X
CLN4 Disease					X
CLN5 Disease			X	X	X
CLN6 Disease			X		X
CLN7 Disease			X		
CLN8 Disease			X	X	
CLN10 Disease	X			X	X
CLN11 Disease					X
CLN12 Disease				X	
CLN13 Disease					X
CLN14 Disease		X			

of not being affected by the disease or being a carrier. When the child develops JNCL, both parents are carriers, and the risk that a sibling will be affected by the same disease is 25 percent. Professional genetic counselling, which includes discussing the various possibilities of prenatal testing, is part of the management for a family where a child has been diagnosed with JNCL.

Disease mechanisms in JNCL

Genes contain information that is necessary for the structure and function of the body. A disease-causing mutation of a gene disables the production of an important molecule, frequently a protein. The lack of this protein then leads to a disturbance of the delicate biochemical network of cells. In the case of JNCL due to a CLN3 defect, the mutation most frequently consists of a large *deletion*, that is, the loss of a piece of the DNA strand that forms the gene. The size of a deletion is measured in kilobase (kb, 1000 base pairs of DNA), and the typical deletion in CLN3 disease has the size of about one kb. Some individuals have smaller mutations, usually

so-called missense or nonsense mutations where a single part of the DNA is not missing but incorrectly sequenced. The symptoms and disease progression of this group may differ somewhat from the majority of individuals with JNCL.

The relevant protein produced under the influence of a non-mutated CLN3 gene is called the CLN3 protein. The size of the CLN3 protein, its chemical structure and location within cells have been characterized and the protein appears to be an important protein because it emerged very early in the evolution of living organisms. It can be detected in a large number of organisms as well as in many different cells of the human body. It is disappointing, therefore, that despite extensive research, the true function of the CLN3 protein has not been fully clarified. It is a structural component of the biological membranes that surround

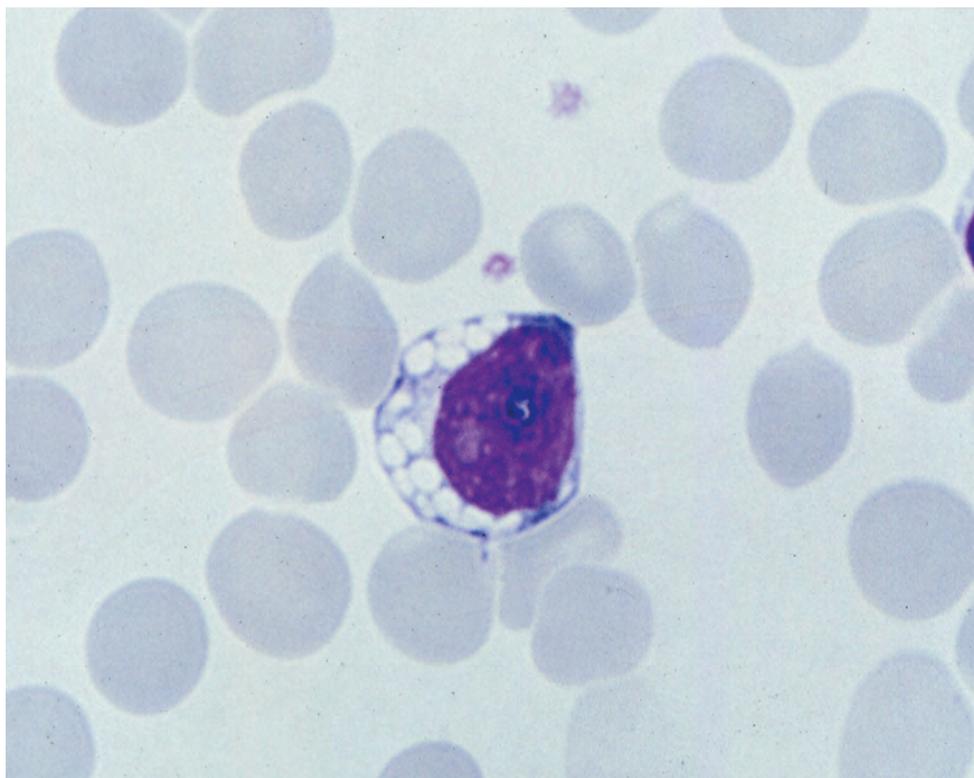


Figure 3.1 Microscopic picture of blood cells from a young person with JNCL

In the center is a single abnormal white blood cell (a lymphocyte). The many vacuoles (white holes) in the cytoplasm (the brighter part of the cell) are evidence of the storage disease. Originally the holes contained stored ceroid lipofuscin, which was lost during preparation for microscopy. The many round grey shadows in the neighborhood of the lymphocyte are normal red blood cells (erythrocytes).

all cells and individual working parts of the cell (and is therefore a transmembrane protein). One such membrane is that of *lysosomes*, small specialized compartments (organelles) within cells. Lysosomes are the organelles responsible for degrading and recycling used material. If lysosomes do not work properly, undegraded biochemical waste builds up inside cells, and the specific function of a cell is impaired. In the case of JNCL, as in other NCL diseases, lysosomes function poorly and become filled with material that should have been degraded, and this material has given the name to these diseases: ceroid lipofuscin. Because of these mechanisms, the NCL diseases belong to the larger group of lysosomal storage disorders.

The abnormal storage material is found in almost all cells of the body, but most cell types apparently do not seem to be disturbed by this process. Figure 3.1 shows a white blood cell that looks abnormal with Swiss cheese-like holes in it, which contain storage material. Among the many cells of the body, however, there are two cell types that are extremely sensitive to the deficiency of the CLN3 protein: light-sensitive cells of the retina and the neuronal cells within the nervous system, mainly in the brain. Although these cells are able to work normally during the first years of life, they work less well later on and eventually die, a process called neurodegeneration. All medical symptoms of JNCL are a consequence of this process, many details of which have not yet been clarified. The general medical hallmark of JNCL, as of most other forms of NCL, is the combination of visual loss through damage to the retina and a progressive brain disease characterized by epilepsy and dementia.

Diagnosis of JNCL

Once JNCL is suspected, making a diagnosis is usually not difficult. However, diagnosis is often delayed for several years after the onset of symptoms; a recently published survey suggested this takes four years on average (Dulz et al., 2015), a delay that has not been much reduced in recent years and is unacceptable. One reason for this delay is the lack of awareness of JNCL among ophthalmologists, who are usually the first to see a child with the beginnings of visual failure. JNCL is the most common cause of severe visual impairment with onset between the age five and fifteen years. Although there are other retinal disorders to consider, a child of school age who begins to have visual problems due to an abnormality of the retina should be suspected of having JNCL until it is proven otherwise.

A first diagnostic test is the microscopic examination of blood cells for the presence of characteristic vacuolated lymphocytes (see Figure 3.1). Technically, this test is relatively simple as it starts with a routine blood smear, but it requires

Table 3.2 Gene defects in the major forms of JNCL

Defective gene	Type of mutation	Remarks
CLN3	Classic deletion (1 kb)	A large majority of individuals with JNCL have this gene defect. The basis of most medical (Åberg et al., 2011) and educational research (von Tetzchner et al., 2013).
CLN3	Missense mutations (may be in combination with the 1 kb deletion)	A rare condition. May have a protracted clinical course (slower progression of the disease than in individuals with classical deletion) or later onset, with only some symptoms (Licchetta et al., 2015)
CLN1	Missense	The disease may start relatively late, visual loss may not be so prominent in the beginning as in individuals with the CLN3 gene (Khan et al., 2013). A potentially treatable enzyme deficiency.
CLN2	Missense	Larger variability of the order in which symptoms appear. Epilepsy may occur early, visual loss may not be the first symptom (Kohan et al., 2012). A treatable enzyme deficiency.

pecially trained personnel who are mainly found only in specialist medical centers. When a high number of lymphocytes with typical vacuoles are found, the diagnosis of JNCL is almost certain. Molecular genetic analysis of the CLN3 gene can be used to detect the most frequent typical deletion in this gene. If the deletion is not present, other mutations in the same gene must be looked for or other genes analyzed, mutations of which can occasionally cause JNCL (see Table 3.2). In most areas, molecular genetic analysis is recommended as the standard procedure for diagnosis.

For the ophthalmologist, the best way to handle the problem is by sending the child to a pediatric medical center with experience in genetic metabolic disorders. Although the necessary tests can be carried out almost anywhere, pediatric experience is desirable for discussing the test results. If results are negative, other rare metabolic or neurological diseases will have to be considered.

A positive test result for JNCL is a matter of great distress for the family. Talking about a disease like JNCL to a family for the first time – preferably to both parents – needs a doctor and other professionals who are adequately informed (Reed et al., 2015). Many follow-up discussions on the nature and prognosis of the disease will be needed with a range of specialists and professionals. Parent organizations provide support and advice through the disease process and it is important to have JNCL teams involved from the outset (see Chapter 9).

Medical treatment of JNCL

JNCL is an incurable disease. For some other forms of NCL that are caused by deficiencies of enzymes, different experimental therapies are in a preclinical stage, while enzyme replacement therapy has already been successful in children with the CLN2 disease (Schulz et al., 2018). JNCL caused by CLN3 mutations is not amenable to such an approach. The reason for this is that the deficient CLN3 protein is not soluble and cannot be replaced in cells with relative ease. Presently such techniques do not work for a protein like CLN3 that is an integral component of membranes.

However, the lack of curative therapy at the present time does not mean that individuals with JNCL cannot be treated for many of the issues arising during the progression of the disease. While most of the treatment options are shared with other neurodegenerative diseases, their application may require a thorough understanding of the disease and how it progresses.

Common problems during disease progression

In view of the characteristic progression of JNCL caused by CLN3 mutations, it has been suggested that the clinical course can be described as a sequence of phases or stages (Kohlschütter et al., 1988; Marshall et al., 2005). There is an overall pattern of loss of functions and abilities, typically with an initial period of mainly visual failure, followed by years with gradually increasing cognitive decline and epilepsy, a growing complexity of problems as motor and language functions decline, and a final phase leading to a comprehensive need for help. However, there is much variability in when cognitive, language and motor problems appear, even in children with the same gene mutation (Lebrun et al., 2011). Moreover, although the phases are typically related to age, they do not always appear at the same time, and the age indications below should be considered approximate. The phases overlap to some degree and the problems caused by the disease may vary considerably. With age, individuals with JNCL may experience a variety of medical problems, including epilepsy, problems with sleep, nutrition and circulation, as well as pain and hallucinations.

Phase 1: Isolated visual failure (4 to 8 years)

During the first years of life, children with JNCL show apparently typical development. The onset of visual problems is usually noted around the age of five to six years, but it often takes several years for a definitive diagnosis to be made (see Chapter 4). One reason may be that ophthalmologists can have difficulties

recognizing that this poor vision is not due to an abnormality of the refractory system of the eye, but of the retina. Examination of the eyes will eventually show severe and characteristic changes (Figure 3.2) but these are less pronounced in this phase of the disease. Electroretinography – a more demanding technical investigation – will early show that the retinas are damaged and do not produce electrical responses when exposed to light (extinguished electroretinogram). Frequently, a misdiagnosis of retinitis pigmentosa is made. This is a disease that leads to blindness but not to the same consequences as a brain disease such as JNCL. Optical coherence tomography (OCT) is a newer non-invasive procedure that will show a characteristically abnormal structure of the retina (Dulz et al., 2015).

Early in development, as vision deteriorates, children with JNCL show a characteristic "overlooking" behavior that can be easily recognized by non-specialists. While the center of the retina is already destroyed, some vision is still possible using the periphery of the retina. So when a child with JNCL is asked to look at you, he or she will often look at a point above or on the side of your head.

The loss of vision tends to occur rather uniformly and rapidly in individuals with JNCL, resulting on average in legal blindness by nine to ten years of age. However, there are noticeable exceptions (see Chapter 4). During this first phase of JNCL, there are usually no other significant medical problems.

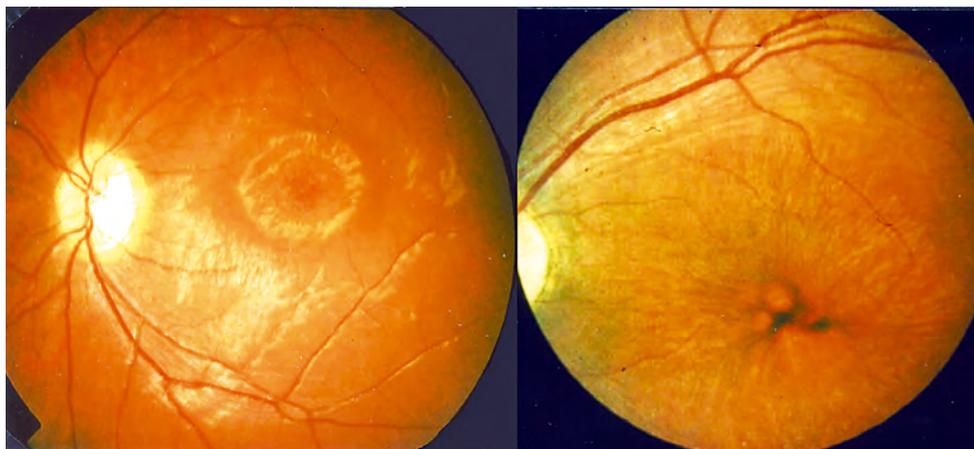


Figure 3.2 Ophthalmologist's view of the light-sensitive retina at the back of the eye in two young people with JNCL

The photograph on the left side shows a "bull's eye" phenomenon in the area of the macula (the spot of greatest visual acuity) in the center, an abnormally pale optic disc (the entrance of the optic nerve) and thinning of blood vessels, indicating decay of the retina. The photograph on the right shows irregular accumulation of blackish pigment, which can lead to the misdiagnosis of retinitis pigmentosa.

Phase 2: Emerging intellectual difficulties (6 to 10 years)

During this phase, it slowly becomes clear that the disease is not restricted to the eyes. Initially, a teacher may observe that a child is still active as normal and has no problems with assignments requiring the use of language, in sports or social communication, but develops unexpected difficulties in mathematics (see Chapter 12). Thinking and talking in particular seem to undergo some changes in children with JNCL in this phase (see Chapters 5 and 6). Eventually, the child will begin to find following lessons so difficult that special educational support and help are required (see Chapter 12). For a while, these difficulties may be attributed to issues of coping with visual loss and may not be regarded as being caused by the dementia process. In some children, behavioral changes (see below) may start in parallel to the cognitive decline.

Phase 3: Increasing medical problems (9 to 14 years)

Epilepsy

A seizure disorder (epilepsy) is a regular component of JNCL (Kohlschütter et al., 2014). The degenerative processes in the brain often lead to abnormal electrical activity or abnormal excitability of the cells during this phase of the disease. The abnormal electrical activity causes muscular convulsions and altered mental states. The first seizure usually occurs at around ten years of age; in the present survey (Appendix A) the first seizure was observed on average at 10.4 years (Sd = 3.0) but with a range from two to nineteen years. Family and teachers should be informed and prepared for this to happen. A number of more or less pronounced types of seizures can occur in JNCL.

A typical and the most dramatic type of seizure is the tonic clonic (used to be called «grand mal») seizure. Without warning, the individual falls, loses consciousness and shows some kind of abnormal movement. The limbs may be stiff (tonic) in the initial phase of the seizure, or move jerkily (clonic). This type of seizure is a combination of the loss of consciousness and unusual motor activity, and implies that the whole brain is in a state of altered electrical activity – a generalized seizure. The stiffness of all muscles of the body during the initial phase of the seizure can hinder normal respiration and lead to a bluish discoloration of lips and skin (cyanosis). After a few minutes, the abnormal movements and the cyanosis subside. The individual falls into a deep sleep, from which he or she awakes with a feeling of exhaustion.

Whilst seizures cause great concern, a generalized tonic-clonic seizure is not usually a life-threatening event. In children and young people with JNCL, such seizures usually last only a few minutes and tend to stop without medical intervention. When a seizure takes place, it helps to turn the person on the side, move items to prevent accidents and observe calmly until the person becomes fully awake and responsive. When a seizure follows this pattern and lasts only a few minutes, emergency transportation to hospital is not usually necessary, but an appointment with the general practitioner or a pediatric neurologist should be made.

Other types of seizures, such as «absence» seizures or «partial» seizures, can be hard to recognize because they may just involve moments of non-responsive behavior or suggest daydreaming. The electroencephalogram (EEG) is a helpful method to determine the presence of subtle seizures and to characterize them.

The propensity of a child or young person for repeated seizures is quite variable. It can be reduced by antiepileptic drugs, but when seizures occur only rarely and can be managed, starting continuous antiepileptic treatment after the first appearance of seizures is not always necessary. The disadvantages of such a treatment needs to be balanced against the desired benefits for each individual. There are excellent antiepileptic drugs to be used in JNCL. As the brain in JNCL is subject to a process of degeneration, the way it reacts to commonly used drugs may not be the same as in children with other conditions (Kohlschütter, Schulz & Denecke, 2014). Some anticonvulsants may be suitable for individuals with JNCL (valproate and lamotrigine), while others may have negative effects on the disease course and should be avoided (carbamazepine, phenytoin, vigabatrin) (Schulz et al., 2013). Some drugs are more prone to produce side effects such as speech disturbance (topiramate) or agitation (levetiracetam and topiramate). Certain drugs used for children with epilepsy may have unusual side effects in individuals with JNCL (Larsen & Østergaard, 2014).

Generally speaking, seizures early in the course of JNCL usually respond well to treatment with antiepileptic drugs, but they will not disappear completely. Later on, seizures may take on a treatment-resistant character. Doctors should bear this in mind and give parents and professionals sufficient information and advice on the possible options so informed choices can be made. Appropriate treatment of epilepsy may be developed with a carefully controlled approach, starting initially with a single drug in low dosage, later using higher dosage. If ineffective, the drug may be changed or combined with another one. A treatment is satisfactory when seizures are suppressed and no side effects present. If this cannot be achieved, it is often better to tolerate some seizures than to experience toxicity from using too many different drugs.

Ideally, the management of a seizure disorder is in the hands of a pediatric neurologist or other clinician who is in contact with an NCL Center. Observations by teachers may be helpful to recognize frequent subtle seizures, which may significantly interfere with the child or young person's attention and thinking. Side effects of antiepileptic drugs, such as excessive sedation, may also be detected at school. With the help of such observations, the dosage of a drug can be modified to provide greater benefits.

Behavioral changes

With time, a child with JNCL realizes that things are different. Apart from the loss of vision, other things that had been previously mastered become more difficult, and the difference in achievements to peers is more noticeable (see Chapter 27). Mood changes, emotional instability and obsessive-compulsive-like behavior (Adams et al., 2006) become noticeable to other people. Frustration can result in depression or aggressive behavior. Help from an experienced child psychologist or psychiatrist may be needed. Poor medical management of epilepsy may add to these problems and should therefore be monitored carefully.

Sleep

A striking observation is the apparent loss of a normal day-and-night rhythm in children with JNCL (Lehwald et al., 2016; Telakivi, Partinen, & Salmi, 1985). Many children and young people with JNCL develop the habit of staying awake until late at night. In the present survey (Appendix A), the average age when sleep problems first appeared was 11 years but variation was large, with a standard deviation of 6.1 years and a range from one to 23 years.

Sleep problems can be disturbing or annoying to the whole family so some form of sleep inducing-medication may be needed. Melatonin is a popular drug in this situation, but does not seem to be very helpful (Hätönen et al., 1999). Alternatives include chloral hydrate, but alterations to a daily routine or lifestyle, as well as a possibly suboptimal antiepileptic treatment should also be considered. Sleep problems may relate to anxiety or other psychological factors (see Chapter 27). Good sleep hygiene should be encouraged. Sometimes a 24-hour EEG is helpful to detect epileptic activity during sleep, as this can add to sleep disturbances and may be managed by improving the anticonvulsive treatment.

Decrease of motor abilities

In late childhood or early adolescence, minor motor problems may become noticeable (see Chapter 7). In this phase, children with JNCL begin to appear clumsy and to lose their balance more easily than previously. Some everyday

activities (dressing, undressing etc.) requiring fine motor abilities may take more time. In a child who has learned braille, these difficulties may interfere with reading.

Phase 4: Puberty, adolescence and young adulthood: Multiple functional losses (15+ years)

Puberty and sex-related issues

Somatic maturation during puberty proceeds without obvious difference. There are some sex-related issues that have not yet been fully clarified. In a large cohort study, the disease showed a slightly more rapid progression in females (Cialone et al., 2012). In another study, several girls were described as having acne and hirsutism, a few had hormonal abnormalities (elevated androgens in blood) and abnormal (polycystic) ovaries. It is possible that some of these abnormalities were related to effects of antiepileptic drugs (Åberg et al., 2002). Menstruation may present problems in personal hygiene because of blindness and decreased mental and fine motor abilities. Girls may experience more frequent and heavier seizures or an excessive increase of tension and nervousness around menstruation. In such situations a gynecologist should be consulted. As well as preventing pregnancy, menstrual suppression – usually by injection of hormones – can be helpful.

Language

In this phase of the disease, verbal communication becomes increasingly difficult due to a combination of problems: cognitive difficulties are increasing and at the same time the motor apparatus required for speaking becomes more severely disturbed. Frustrated by a willingness to communicate but not being able to talk as easily as before, young people with JNCL may develop a peculiar and characteristic way of speaking. Their expressive language becomes more simple in content, it sounds soft and slurred, and often becomes understandable only to persons who are familiar with the person with JNCL (see Chapter 6). Some young people with JNCL appear to be in a hurry to get their idea over to their partner. Their speech contains many repetitive elements, stuttering or repeating of words or short sentences, and may include apparently meaningless phrases. The age at which language becomes hardly understandable is very variable, ranging from eight years to more than 20 years (Lebrun et al., 2011). Comprehension of language persists longer than expressive speech, which may contribute to frustration and aggressive behavior, as a reaction to not being understood. There are several approaches to help both young persons with JNCL and their caregivers to communicate better (see Chapter 13).



Figure 3.3 Typical posture of an adolescent with JNCL

Major motor problems, loss of ambulation

Starting at the age of 10 to 12 years, some children with JNCL develop a peculiar «extrapyramidal» motor disorder that resembles Parkinson disease. In others, this arises later on (Järvelä et al., 1997). Symptoms of this disorder are hypokinesia (decreased bodily movement), rigidity (stiffness) of muscles, stooped posture (see Figure 3.3), shuffling gait and impaired balance. The symptoms can sometimes be temporarily ameliorated by anti-Parkinson medication (dopa or similar drugs) (Åberg et al., 2001). Some young people with JNCL become wheelchair-bound at around 16 years of age, but the age when this happens is very variable (from 11 to 26 years) (Lebrun et al., 2011).

Decrease in intellectual abilities

Visual loss and speech impairment may make cognitive assessment difficult beyond the age of 15, but dementia may become more severe and include decline in memory, attention, emotional control, and general reasoning abilities (see Chapter 5). There is also likely to be a decline in independence skills like mobility, feeding and communication. The need for support among individuals with

JNCL can be likened to the care needed by older people with Alzheimer disease or other forms of dementia. The aim of support in this area is to provide reassurance to the young persons and their relatives, and to avoid that they become isolated and inactive.

Behavioral, psychological and psychiatric issues

Children and young people with JNCL may show many behavioral problems and psychiatric symptoms (Adams et al., 2006; see Chapter 27). In a group of individuals with JNCL aged between 9 to 21, the most common symptoms reported by parents and other caregivers were problems with social interaction, thinking, attention, aggressive behavior and some less well defined somatic complaints

(Bäckman et al., 2005), as well as anxiety and depression. While similar problems are frequent in individuals with intellectual disability (Myrbakk & von Tetzchner, 2008), they are often recognized late in young people with JNCL and treatment is often insufficient. Hallucinations are frequent in the late phase of JNCL (Lanska & Lanska, 1993). A hallucination is a vivid experience of something that appears to be located in external objective space, typically an episode of several minutes duration, and perceived by the subject as real. It is important for caregivers to be aware of the individual's sense of reality of the experience (see Chapter 25). Hallucinations may be harmless, even pleasant. The individual may be pausing without apparent motive or movement, or may smile without any comment. Quite different and potentially dramatic are hallucinations with an unpleasant content. A hallucinating person may have the subjective experience that the house is on fire, he or she might say (or think) that immediate action is required or try to run away. When this happens in the classroom and does not stop spontaneously, it can be distressing for all involved. Reassuring words like «Look, nothing feels hot in the room, there is no smoke,» may not be sufficient to calm the person down. The situation may develop into a psychiatric emergency and require the use of strong sedative medication and further psychiatric management.

In this phase of the disease, young people with JNCL are generally less able to tolerate a change in their environment. They feel safer, more comfortable and show more emotional stability in familiar environments and in the presence of people they know. Such considerations may be of value when decisions relating to education and accommodation have to be made.

Other somatic problems

Nutrition

There is no need for a special diet. Constipation should be avoided, as this can become a chronic problem in later stages of the disease.

Circulation

Children and young people with JNCL frequently have cold hands and feet without abnormal findings when examined. Later in the disease progression, the heart may be affected (Østergaard, Rasmussen, & Mølgaard, 2011), primarily resulting in a slowing of the heartbeat. In exceptional cases, late in the course of the disease, surgical insertion of a cardiac pacemaker can significantly improve well-being. In adolescents, a specific treatment for a heart condition is not usually necessary, but the heart should be checked as part of the medical follow-up.

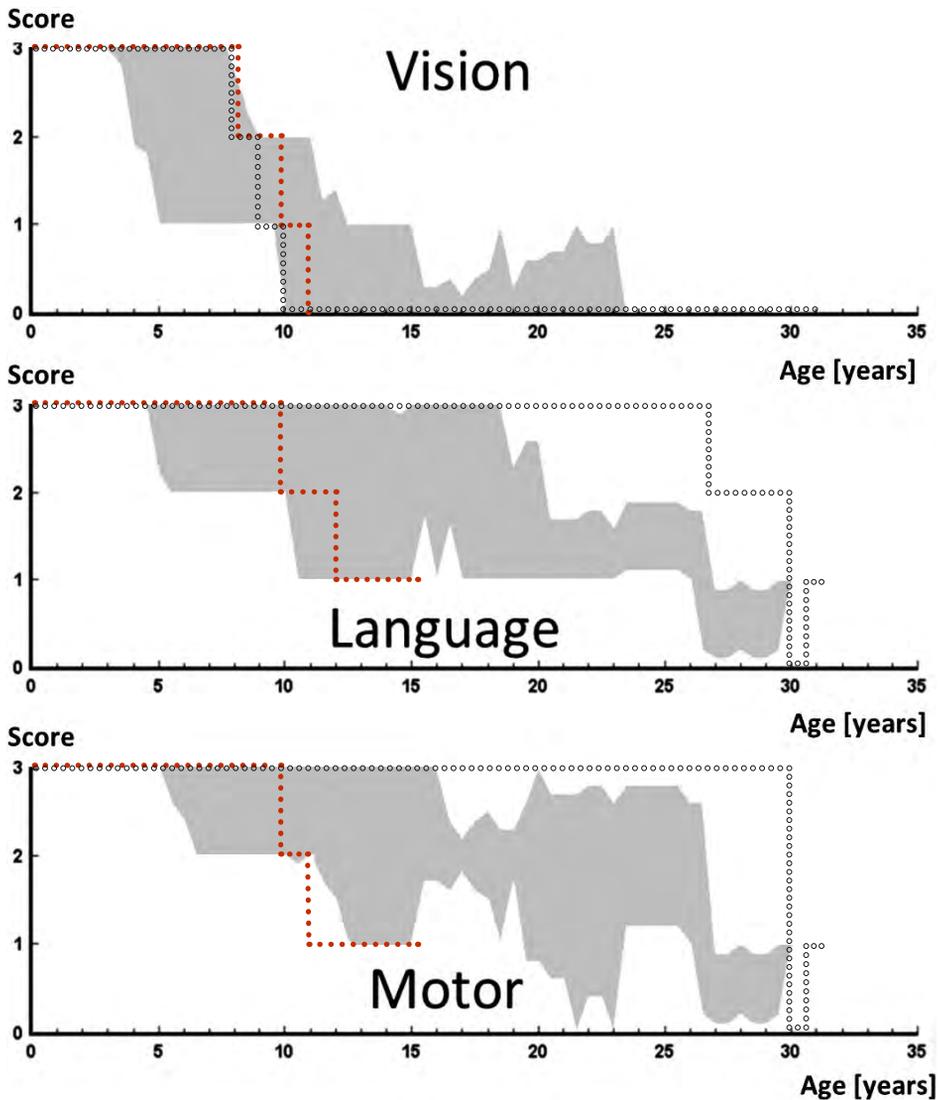


Figure 3.4 Results from a study of 25 individuals with JNCL with identical mutations (classical deletion) in the *CLN3* gene

Scoring: (3) normal function, (2) slight problems, but readily recognized, (1) severe problems, and (0) total loss of function. The shaded area represents scores between the 10th and 90th percentile. The diagram illustrates that the vision impairment occurs quite uniformly, while the loss of language and motor functions is more variable (Lebrun et al., 2011, p. 1255). The dotted red lines represent the course in an individual with a particularly fast loss of language and motor function, while the line of black circles represents the course in an individual with particularly slow loss of both functions. The second individual showed some improvement in motor and language functions after the implantation of a cardiac pacemaker at age 30 years.

Pain

When a young person with JNCL has lost the ability to communicate successfully, any situation where he or she expresses unhappiness or pain without apparent cause, raises difficult questions. When the problem does not subside spontaneously, a doctor should look for possible abnormalities in organs before analgesics are prescribed.

Concluding comments

Following the individual trajectories of JNCL over many years offers a striking observation: while the loss of vision proceeds quite uniformly and is very similar in all individuals, the decline of language and motor abilities take a more variable course with large individual variation (Figure 3.4). The reasons for this are not known, nor why there are differences between typical courses in males and females. Understanding the reasons for this might help to improve medical treatment. It cannot be ruled out that variable quality of the medical treatments used may contribute to these differences. It is therefore very important to carefully monitor treatments and the disease progression in all children and young people with JNCL throughout their life.

There are further medical issues that can arise during the disease progression that are not addressed within the scope of this book. Families should know that specific medical experience with JNCL has developed so that whatever their child may experience, expert advice is available (see Appendix B).

Acknowledgments

We are grateful to all the families who completed the survey, for discussions with members of Bildungszentrum für Blinde und Sehbehinderte in Hamburg and Bartiméus Foundation in Doorn, the Netherlands. Verein der Freunde der Kinderklinik UKE e.V. has generously supported the Clinic for Children with Degenerative Brain Diseases in Hamburg.

References

- Åberg, L. E., Rinne, J. O., Rajantie, I., & Santavuori, P. (2001). A favorable response to antiparkinsonian treatment in juvenile neuronal ceroid lipofuscinosis. *Neurology*, *56*, 1236–1239.
- Åberg, L. E., Tiitinen, A., Autti, T. H., Kivisaari, L., & Santavuori, P. R. (2002). Hyperandrogenism in girls with juvenile neuronal ceroid lipofuscinosis. *European Journal of Paediatric Neurology*, *6*, 199–205.

- Åberg, L., Autti, T., Braulke, T., Cooper, J. D., van Diggelen, O. P., Jalanko, A., et al. (2011). In S. E. Mole, R.E. Williams, & H. H. Goebel (Eds.), *The neuronal ceroid lipofuscinoses (Batten disease)* (pp. 110–139). Oxford, UK: Oxford University Press.
- Adams, H., de Blicq, E. A., Mink, J. W., Marshall, F. J., Kwon, J., Dure, L., Rothberg, P. G., Ramirez-Montealegre, D., & Pearce, D. A. (2006). Standardized assessment of behavior and adaptive living skills in juvenile neuronal ceroid lipofuscinosis. *Developmental Medicine and Child Neurology*, *48*, 259–264.
- Bäckman, M. L., Santavuori, P. R., Åberg, L. E., & Aronen, E. T. (2005). Psychiatric symptoms of children and adolescents with juvenile neuronal ceroid lipofuscinosis. *Journal of Intellectual Disability Research*, *49*, 25–32.
- Cialone, J., Adams, H., Augustine, E. F., Marshall, F. J., Kwon, J. M., Newhouse, N., et al. (2012). Females experience a more severe disease course in batten disease. *Journal of Inherited Metabolic Disease*, *35*, 549–555.
- Dulz, S., Wagenfeld, L., Nickel, M., Richard, G., Schwartz, R., Bartsch, U., Kohlschütter, A., & Schulz, A. (2015). Novel morphological macular findings in juvenile CLN3 disease. *British Journal of Ophthalmology*, *100*, 824–828.
- Hätönen, T., Kirveskari, E., Heiskala, H., Sainio, K., Laakso, M. L., & Santavuori, P. (1999). Melatonin ineffective in neuronal ceroid lipofuscinosis patients with fragmented or normal motor activity rhythms recorded by wrist actigraphy. *Molecular Genetics and Metabolism*, *66*, 401–406.
- Järvelä, I., Autti, T., Lamminranta, S., Åberg, L., Raininko, R., & Santavuori, P. (1997). Clinical and magnetic resonance imaging findings in batten disease: Analysis of the major mutation (1.02-kb deletion). *Annals of Neurology*, *42*, 799–802.
- Khan, A., Chieng, K. S., Baheerathan, A., Hussain, N., & Gosalakkal, J. (2013). Novel CLN1 mutation with atypical juvenile neuronal ceroid lipofuscinosis. *Journal of Pediatric Neurosciences*, *8*, 49–51.
- Kohan, R., Carabelos, M. N., Xin, W., Sims, K., Guelbert, N., Cisondi, I. A., et al. (2013). Neuronal ceroid lipofuscinosis type CLN2: A new rationale for the construction of phenotypic subgroups based on a survey of 25 cases in South America. *Gene*, *516*, 114–121.
- Kohlschütter, A., Laabs, R., & Albani, M. (1988). Juvenile neuronal ceroid lipofuscinosis (JNCL): Quantitative description of its clinical variability. *Acta Paediatrica*, *77*, 867–872.
- Kohlschütter, A., Schulz, A., & Denecke, J. (2014). *Epilepsy in neuronal ceroid lipofuscinoses (NCL)*. *Journal of Pediatric Epilepsy*, *3*, 199–210.
- Lanska, D. J., & Lanska, M. J. (1993). Visual «release» hallucinations in juvenile neuronal ceroid-lipofuscinosis. *Pediatric Neurology*, *9*, 316–317.
- Larsen, E. P., & Østergaard, J. R. (2014). Valproate-induced hyperammonemia in juvenile ceroid lipofuscinosis (Batten disease). *Seizure – European Journal of Epilepsy*, *23*, 429–434.
- Lebrun, A. H., Moll-Khosrawi, P., Pohl, S., Makrypidi, G., Storch, S., Kilian, D., et al. (2011). Analysis of potential biomarkers and modifier genes affecting the clinical course of CLN3 disease. *Molecular Medicine*, *17*, 1253–1261.
- Lehwald, L. M., Pappa, R., Steward, S., & de los Reyes, E. (2016). Neuronal ceroid lipofuscinosis and associated sleep abnormalities. *Pediatric Neurology*, *59*, 30–35.
- Licchetta, L., Bisulli, F., Fietz, M., Valentino, M. L., Morbin, M., Mostacci, B., et al., (2015). A novel mutation of CLN3 associated with delayed-classic juvenile ceroid lipofuscinosis and autophagic vacuolar myopathy. *European Journal of Medical Genetics*, *58*, 540–544.

- Marshall, F. J., de Blicke, E. A., Mink, J. W., Dure, L., Adams, H., Messing, S., et al. (2005). A clinical rating scale for Batten disease reliable and relevant for clinical trials. *Neurology*, *65*, 275–279.
- Myrbakk, E., & von Tetzchner, S. (2008). Psychiatric disorders and behavior problems in people with intellectual disability. *Research in Developmental Disabilities*, *29*, 316–332.
- Østergaard, J. R., Rasmussen, T. B., & Mølgaard, H. (2011). Cardiac involvement in juvenile neuronal ceroid lipofuscinosis (Batten disease). *Neurology*, *76*, 1245–1251.
- Reed, S., Kassis, K., Nagel, R., Verbeck, N., Mahan, J. D., & Shell, R. (2015). Breaking bad news is a teachable skill in pediatric residents: A feasibility study of an educational intervention. *Patient Education and Counseling*, *98*, 748–752.
- Schoenberg, M. R., & J.G. Scott, J. G. (2011). Cognitive decline in childhood or young adulthood. In M. R. Schoenberg & J. G. Scott (Eds.), *The little black book of neuropsychology: A syndrome-based approach* (pp. 839–861). New York: Springer.
- Schulz, A., Ajayi, T., Specchio, N., de Los Reyes, E., Gissen, P., Ballon, D., et al. (2018). Study of intraventricular cerliponase alfa for CLN2 disease. *New England Journal of Medicine*, *378*, 1898–1907.
- Schulz, A., Kohlschütter, A., Mink, J., Simonati, A., & Williams, R. (2013). NCL diseases—clinical perspectives. *Biochimica et Biophysica Acta (BBA)-Molecular Basis of Disease*, *1832*, 1801–1806.
- Stengel, C. (1826). Beretning om et mærkeligt Sygdomstilfælde hos fire Sødskenne i Nærheden af Røraas (*The history of a strange case of disease in four siblings near Røros*). *Eyr et medicinsk Tidsskrift*, *1*, 347–352.
- Telakivi, T., Partinen, M., & Salmi, T. (1985). Sleep disturbance in patients with juvenile neuronal ceroid lipofuscinosis: A new application of the SCSB-method. *Journal of Intellectual Disability Research*, *29*, 29–35.
- von Tetzchner, S., Fosse, P., & Elmerskog B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta*, *1832*, 1894–1905.
- Williams, R. E., & Mole, S. E. (2012). New nomenclature and classification scheme for the neuronal ceroid lipofuscinoses. *Neurology*, *79*, 183–191.

4

Development and Loss of Vision

Bengt Elmerskog and Stephen von Tetzchner

Perception is the ability to distinguish and identify sensory information, to direct and sustain attention to various aspects of the environment, and to lend meaning to them. Children use their senses to explore the physical and social world, establish a basis for action, and to monitor and regulate their own actions. The sensory systems facilitate interaction with other people. Vision is the sense that provides the most information about the environment (von Tetzchner, 2019).

Visual impairment

Visual impairment is defined as a reduced ability to perceive and interpret visual sensory stimulation beyond what may be compensated with glasses. The impairment may consist in reduced visual acuity or field of vision, or a combination of these two functions. Visual impairment ranges from "normal or mild" to "(total) blindness" (Table 4.1). Normal vision is usually described as 6/6, meaning that the person with best correction (glasses) at a distance of six meters can see the same as most people can at that distance. Moderate visual impairment ranges from 6/18 to 6/60, indicating that the person only at a distance of six meters can see what people with normal sight can see at 18 to 60 meters. The British legal definition of blindness is 3/60, which means that the person needs to be at a distance of three meters to be able to see what people with normal sight can see at 60 meters. The US definition is 6/60. "Near total blindness" indicates some light perception while "total blindness" implies that the person has no light perception (World Health Organization, 2010).

Mild visual impairment is common and can in many cases be corrected with glasses (Wiener, Welsh, & Blasch, 2010). Approximately 1–2 percent of school children have reduced vision with functional consequences. Many children with severe visual impairment have additional disorders, particularly in mobility,

Table 4.1 Categories of visual impairment

Categories of visual impairment		Worse than	Equal or better than
0	Mild or no visual impairment		6/18
1	Moderate visual impairment	6/19	6/60
2	Severe visual impairment	6/60	3/60
3	Blindness	3/60	1/60
4	Blindness (near total)	1/60	Light perception
5	Blindness (total)	No light perception	

Note: Degree of visual impairment is described as degree of vision related to typical vision. Average normal vision is usually described as 6/6, meaning that the person can see the same at a distance of six meters as most people do. Moderate visual impairment ranges from 6/19 to 6/60, indicating that the person can see at six meters what people with normal vision can see at 19 to 60 meters.

hearing impairment, intellectual disability (Geddie et al., 2013). Although rare, JNCL continues to be the most common cause of blindness in many countries in children aged 5–15 years (Mole et al., 2011). There is much research on the development of children with congenital or early acquired blindness. Less is known about the developmental consequences of blindness acquired in middle childhood (age 6–12 years).

Consequences of visual impairment

The visual sense is so basic to human functioning that severe visual impairment will cause difficulties in many areas of development. Many ordinary activities of daily living become difficult to master, such as pouring liquid into a cup, finding things and orientating in the physical environment (Brambring, 2007; Dale & Edwards, 2015; Fraiberg, 1977). For people with low vision, access to visual information may be enhanced with technical aids, improved lighting, and magnification, whereas individuals with blindness often must learn to do things in a different way to cope in the environment (see Chapter 16). Even a minimum of light perception may make it easier for the person to acquire many daily living skills.

Early visual impairment will typically affect motor development and physical status (see Chapter 7), and children who are visually impaired may be less physically fit than their sighted peers (Aslan, Calik, & Kitis, 2012; Skaggs & Hopper, 1996).

Independence in activities of daily living may be challenging for most individuals with severe visual impairment, particularly those activities involving movement from one location to another. An early- or late-acquired blindness

will always entail an increased dependence on others, as well as special needs for learning and physical adaptations (Wiener et al., 2010). For instance, for persons with total blindness, orientation in space may depend on stable placement of objects and of topographic patterns in the surroundings that can be detected by touch or hearing (see Chapter 16).

Most ordinary social activities for children require vision. Severe visual impairment will restrict possibilities for participating in soccer, bicycling, window shopping, and so forth. Although many children walk or bicycle to school, children who are blind may need to go to school by car or be accompanied by an adult. Children with normal sight may explore their surroundings visually on their way from a starting point to a destination. Children's movements, like play, can exist for their own sake without the need for some goal (e.g., reaching a destination). Thus, like play, they constitute important learning elements in children's development (Bodrova and Leong, 2015; Lillard, 2001; Piaget, 1951). In most cases, children who are blind face barriers to initiating and participating in such activities and there may be consequences for their overall development (Fraiberg, 1977).

Metaphorically, Gibson (1979) describes perception of the environment in a person who is blind as a "bubble" or "tunnel", while a person with normal sight perceives the environment as a "field", "spectrum" or "map". Further, sighted people can perceive spatial information in a single glance, while individuals who are blind need to explore, synthesize, reconstruct and memorize to obtain similar information about the physical environment.

Visual information may to some extent be compensated with auditory and tactile information. Tactile information, for example about an object landmark, depends on the object of investigation being reachable by hands or body. For people who are blind, orientation in space therefore requires directed attention and more cognitive resources, and patterns or structures in the environment that enable them to move independently (see Chapter 16). Independent goal-oriented movement from place to place requires anticipation of learned landmarks and an understanding of environmental features that cannot be perceived with touch or hearing. Knowing where one is in physical space, where objects are in relation to each other and to self, and how these relations change with self-movement, represent life-long challenges for people who are blind (Tellevik, Storliløkken, Martinsen, & Elmerskog, 1998; Wiener et al., 2010).

Moreover, the independence and autonomy of the individual may be negatively influenced when caregivers, in an effort to help or to rush through activities, complete tasks for the individual with blindness, sometimes leading to learned helplessness (Dodds et al., 1991). Such behaviors from caregivers may hinder efforts of individuals who are blind to learn about the world through the remaining senses, particularly hearing and touch (Willings, 2017).

For persons who are blind, independent living, without assistance from sighted helpers, is limited to familiar environments; however, independence outside these areas will almost always involve support from sighted helpers. People who are blind therefore need to learn (and dare to and are motivated to learn) how they can use sighted people in the surroundings as tools to achieve personal goals. Some authors define such self-governed strategies as "interdependence" (Tellevik & Elmerskog, 2001; see Chapter 16).

There is evidence that people who are blind may develop stronger memory skills that compensate for the absence of vision in developing and maintaining a social and independent lifestyle (Withagen, Kappers, Vervloed, Knoors, & Verhoeven, 2013). For instance, features in the environment that cannot be perceived by vision, owing to the impairment, must be mapped and memorized using information from the other senses, like touch or hearing. Such behaviors require extensive and time-consuming exploration and memory (see Chapter 16).

Early and later vision loss

The consequences of visual impairment depend on age of onset of visual loss. Visual impairment that is present from birth (congenital) will have a greater impact on development and learning than visual impairment acquired later. A great deal of an infant's learning about the world is through the visual modality. Learning by observation is considered an important way of learning but is limited for children with congenital severe visual impairment. Social development is also affected, because children who are blind are not able to pick up on non-verbal cues or make eye contact. In addition, children who are blind may appear disinterested (their attentive mode may look different from that of sighted peers), resulting in reduced sustained social interactions with peers (Perez-Pereira & Conti-Ramsden, 1999).

The surroundings will in many cases appear less stimulating for children who are blind compared to children with sight (Adelson & Fraiberg, 1974). Loss of vision will thus have a negative impact on motor development because the child is lacking the visual incentive to move toward things, and this deficit may cause inhibition to move for fear of the unknown (Brambring, 2006; Freeman & Cannady; 1971; Levtzion-Korach et al., 2000). Getting to know the environment is essential for cognitive development, and exploration requires both the ability to move and an incentive to move towards particular objects and people. Without vision there may be little incentive to explore things that require close inspection and contact and cannot be appreciated from afar. Thus, reduced exploration may influence both motor development and conceptual development.

Children with congenital blindness have little or no visual experience, and so they build up their understanding of the world from non-visual experiences.

Children with later onset of visual impairment, like children with JNCL, have already learned about the presence of people and objects in the environment and how to use vision to orientate in the world. Unlike children with congenital blindness, they have acquired typical everyday skills in the same way as most children and hence have a different basis for learning new skills later in their development.

The role of hearing in visual impairment

Hearing has the same functions in blind as in sighted children, but in addition hearing to some extent compensates for visual loss (Elmerskog, Martinsen, Storliløkken, & Tellevik, 1993). Getting to know the immediate environment and being able to orientate in the physical surroundings are core human functions. In the development of mental spatial representations, sighted children use geometric cues and landmarks. Geometric cues include surface, direction, distance, angle and the like, which depend on the viewer's perspective (egocentric cue). Landmarks are external cues and include characteristics such as wall color and placement of windows, doors and objects, and are independent of viewer location (Ferrara & Landau, 2015). Children who are blind need more time to explore and form mental representations of space based on their experiences with locomotion, non-visual strategies and landmarks, and the need for time may have consequences for their development of exploration and independence (Schmuckler & Gibson, 1989; McAllister and Gray, 2007). Individuals with congenital blindness use hearing to develop mental spatial representations from sounds, but their representations may be based more on egocentric cues – anchored in own body – while sighted individuals proceed to use more external cues (Vercillo, Tonelli, & Gori, 2017). Although hearing provides important information about the direction from which a sound is coming, it gives less precise information about its distance from the listener, and this influences the spatial map of individuals with congenital blindness (Kolarik et al., 2017). Despite the ability to hear, acquired blindness will cause major changes in the individual's life and limit personal perspectives and control of the surroundings.

Some children with congenital blindness develop outstanding listening skills, such as echolocation and perfect pitch. Such abilities are most prominent in individuals with onset of visual problems in early childhood (Dingfelder, 2005; Wan et al., 2010). Echolocation is the ability to detect location of objects by sensing echoes from stomping the foot, snapping the fingers, or making clicking sounds. Children who are blind can identify objects' location, size, and possibly even material composition from a relatively small distance by using echolocation. Good skills in echolocation make the orientation skills of people

who are blind more efficient, and these skills are developed through active and independent exploration (Elmerskog et al., 1993). In addition, verbal explanations and descriptions about things and events in the surrounding can provide richer, supplementary information. Yet despite these rich compensations, most information that may be available through vision will not be available to persons who are blind. Specifically, sighted people are typically using visual references in communication, which may not be useful for people without sight. For instance, it may be useful to apply egocentric references (relative to the position of the person) rather than topographic references in communication with persons without sight (see Chapter 16).

Moreover, auditory information gives a different conception of the environment than vision. Vision gives much information simultaneously, provided there is enough light, including information about the current spatial relationship of things and people in the environment, and how they move. Many objects do not make sounds and may be out of the perceptual field, inaudible, most of the time. Perceived stability will depend on the sound being continuous or repeated. Localization and identification of a sound are two separate processes in hearing (Gibson, 1966). For instance, a scratchy sound created by a person moving on a graveled road can be identified and localized at the same time for a person with vision. A person who is blind can localize the sound through hearing but may not be able to determine the source of the sound unless it is recognized as a sound that has been heard before.

Many people who are blind show exceptional skill in using sound to navigate in space and explore the physical and social environment. However, in some ways, the information provided by vision or by sound may create somewhat different impressions of the environment.

Visual impairment in individuals with JNCL

The first symptom of JNCL is typically visual loss due to retinal degeneration between four to ten years of age (see Chapter 3). Children with JNCL show decreasing visual acuity and reduced luminance and chromatic contrast sensitivity. They often have large central retinal visual field defects (scotoma) and may compensate for these by using para-foveal or para-macular fixation techniques. This means that the child uses part of the peripheral visual field rather than the central part, for example by fixating above, below or next to the natural foveal fixation point (Augestad, Fosse, & Didrichsen, 2008). Most of the participants in the present study (see Appendix A) showed normal development prior to onset of visual problems. A few parents reported some cognitive or behavioral

problems prior to the visual impairment, but none of these symptoms led to a JNCL diagnosis.

In the present survey, parents had first become aware of their child's visual problems at an average age of 6.2 years (SD 1.7, range 2-12) (see Figure 4.1). A little later, at around 7.8 years (SD 2.0, range 4-14), the visual impairment began to have a major impact on everyday functioning. Around the age of 10.7 (SD 3.4, range 5-20), the child had become blind. The average time period from when the first symptoms emerged to when the children had become blind was rather short, around 4½ years. However, there was still considerable variation in the following variables: (a) age at onset of visual problems, (b) age when these problems made a significant impact, and (c) age when the child became blind; and there were exceptional cases with much earlier or later age of onset. Despite the rather short average time span, the results indicate a larger variation in visual decline among children with JNCL than has usually been assumed. This finding is important when starting to plan for the education and environmental adaptation for a child in this group. Many children with JNCL utilize their vision actively for orientation and in leisure activities throughout the early school years, and they may use residual vision effectively for learning purposes until the age of 9–10 years, in some cases well into the teens or even after the age of 20 (see Chapter 12).

Children with JNCL have several years of visual experience when the visual decline starts. The development in this period constitutes a foundation for how they think and act in the world. Children with JNCL know what the world looks like, they have developed an understanding of spatial concepts and space based on visual experiences, they know what colors are, and they have developed

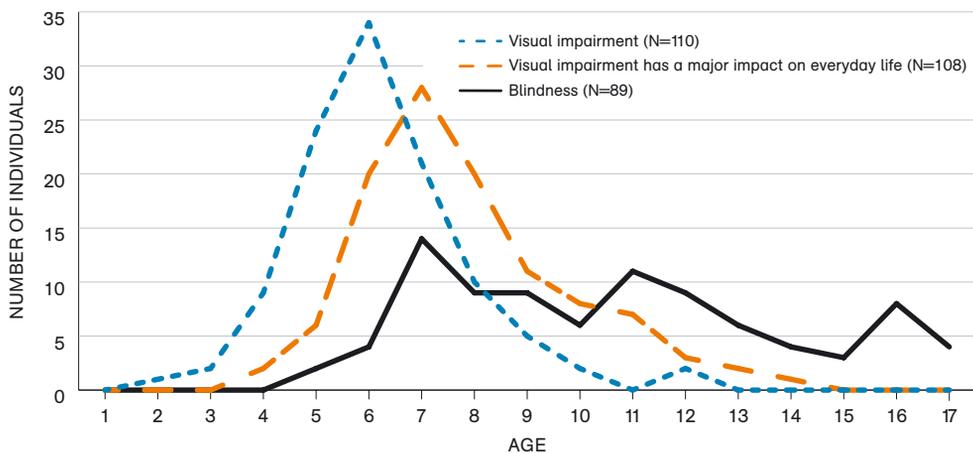


Figure 4.1 Visual decline in JNCL

interests and preferences in a similar manner as their peers. The content of their conversation after the onset of blindness illustrates the continuous role of their visual experiences. Colors of clothes and dolls were important for many children after the onset of blindness, they were using visual references and concepts when describing the environment, and they used gestures and other vision-dependent non-verbal expressions to underline their own meanings like sighted children do. In fact, in the present survey some parents and staff reported that the visual references based on memories were so pervasive that the children and young people with JNCL believed that they had seen objects or events after they had been diagnosed with total blindness. Moreover, there were no reports of the use of echolocation or other outstanding auditory abilities often observed in children with congenital blindness.

The results further emphasize the difference in functioning between children with congenital blindness and children with JNCL. Almost no children with JNCL showed problems with passivity, self-stimulation or self-understanding in early life as do children with congenital blindness. However, many parents found that their child's life became more passive after he or she became blind, and this was more prominent after the age of 16, several years after the onset of blindness. This may suggest that problems with passivity were not primarily caused by the visual impairment. Emerging dementia and motor problems, as well as reduced access to activities with friends, may have contributed to the development of a more passive life style (see Chapter 21).

Individuals with JNCL and other forms of early acquired visual impairment must learn how to cope with the world in new ways. This coping requires mental and cognitive effort, and it may be easier for children to begin learning how to cope while their cognitive capacity and ability to learn are as good as possible (see Chapters 12 and 16). The difficulty lies in the finding that cognitive decline (dementia) in children with JNCL evolves not long after the emergence of severe visual impairment (see Chapter 5). Cognitive abilities and difficulties are important considerations for children's education. However, the findings of the present survey suggest that to some extent, the visual impairment seemed to over-shadow possible cognitive problems in the early years, and cognitive problems received limited attention from the schools and educational authorities, or even from JNCL counselors. In fact, cognitive and visual decline seemed rather to be approached as two discrete, unrelated phenomena leading to fragmented intervention measures.

Of special importance is how people in the surroundings respond to the child's emergent visual problems and special needs. A compassionate response may result in over-eagerness to help or assist the child in many situations, to do things for the child that the child is able to do herself, or to wait with useful educational initiatives until the situation has "calmed down". It is important to

emphasize that besides the gradually increasing visual problems, most children with JNCL do not experience big changes in connection with getting the diagnosis. Their life goes on as usual and most children in the present study were not aware of their own diagnosis at this point in time. However, the child's diagnosis will have a major impact on parents and others who are informed about it.

We experienced that we received a new son after the diagnosis was delivered to us. Our old son with all expectations about the future were gone in a glance and replaced with a new son with alternative expectations for the future.

Reactions from people the environment, transactionally caused partly by the child's visual problems and changing behavior (see Chapter 2), may impose an indirect impact on the child's everyday life (see Chapters 22 and 27). Teachers and other professionals may also show exaggerated compassionate reactions. It is a risk that such reactions will interfere or impede the development or maintenance of independence in young persons with JNCL. For instance, a child or adolescent with JNCL who originally had a high degree of independence, may become accustomed to things being done on his or her behalf. However, it may support maintenance of independence and autonomy when young people with JNCL learn to use sighted people as tools to achieve personal goals in unfamiliar environments. Such self-governed strategies may be defined as interdependence (see Chapter 16).

Conclusions

Visual loss is one of the defining symptoms of JNCL and the visual problems that typically begin to emerge in early childhood are in most cases the main basis for the diagnostic process. There is considerable research on the development of children with congenital blindness, and this research represents one important source for understanding the development of children with JNCL. Nevertheless, there are significant differences between the development of children with congenital blindness or onset of blindness in infancy, and the development of children with gradually increasing visual impairment later in childhood. In addition, the visual problems interact with the emergent cognitive problems in the development of children with JNCL. A necessary basis for goal-oriented and adapted education is an understanding of how the visual problems work together with the cognitive problems in development, learning and participation. In many cases, adapted education will necessitate inclusion of an extended curriculum for children and adolescents with JNCL.

References

- Adelson, E., & Fraiberg, S. (1974). Gross motor development in infants blind from birth. *Child Development, 45*, 114–126.
- Aslan, U. B., Calik, B. B., & Kitiş, A. (2012). The effect of gender and level of vision on the physical activity level of children and adolescents with visual impairment. *Research in Developmental Disabilities, 33*, 1799–1804.
- Augestad, L. B., P. Fosse, P., & Diderichsen, J. (2008). Symptomatisk behandling av pasienter med barndomsformer av Nevronal Ceroid Lipofuscinose (NCL) (Symptom management of patients with the juvenile form of NCL). *Tidsskrift for Norsk Psykologforening, 45*, 33–38.
- Bodrova, E., & Leong, D. J. (2015). Vygotskian and post-Vygotskian views on children's play. *American Journal of Play, 7*, 371–388.
- Brambring, M. (2006). Divergent development of gross motor skills in children who are blind or sighted. *Journal of Visual Impairment and Blindness, 100*, 620–634.
- Brambring, M. (2007). Divergent development of manual skills in children who are blind or sighted. *Journal of Visual Impairment and Blindness, 101*, 212–225.
- Dale, N., & Edwards, L. (2015). Children with specific sensory impairments. In A. Thapar, D. S. Pine, J. F. Leckman, S. Scott, M. J. Snowling and E. Taylor (Eds.), *Rutter's child and adolescent psychiatry*, Sixth edition (pp. 612–622). Chichester, West Sussex, UK: Wiley.
- De Boer, J., Wulffraat, N., & Rothova, A. (2003). Visual loss in uveitis of childhood. *British Journal of Ophthalmology, 87*, 879–884.
- Dingfelder, S. (2005). Pitch perfect. Everyone may be able to learn to name pitches, but the window of time to do it occurs only early in life. *Monitor on Psychology, 36* (2).
- Dodds, A. G., Bailey, P., Pearson, A., & Yates, L. (1991). Psychological factors in acquired visual impairment: The development of a scale of adjustment. *Journal of Visual Impairment and Blindness, 85*, 306–310.
- Dulz, S., Wagenfeld, L., Nickel, M., Richard, G., Schwartz, R., Bartsch, U., Kohlschütter, A. & Schulz, A. (2016). Novel morphological macular findings in juvenile CLN3 disease. *British Journal of Ophthalmology, 100*, 824–828.
- Elmerskog, B., Martinsen, H., Storliløkken, M., & Tellevik, J. M. (1993). *Førlighetsopplæring – Mobilitet i en funksjonell sammenheng* (Mobility training in a functional context). Trondheim: Tapir.
- Ferrara, K., & Landau, B. (2015). Geometric and featural systems, separable and combined: Evidence from reorientation in people with Williams syndrome. *Cognition, 144*, 123–133.
- Fraiberg, S. (1977). *Insights from the blind*. New York: Basic Books.
- Fraiberg, S., & Freedman, D. A. (1964). Studies in the ego development of the congenitally blind child. *The Psychoanalytic Study of the Child, 19*, 113–169.
- Freedman, D. A., & Cannady, C. (1971). Delayed emergence of prone locomotion. *Journal of Nervous and Mental Disease, 153*, 108–117.
- Freeman, R. D., Goetz, E., Richards, D. P., & Groenveld, M. (1991). Defiers of negative prediction: A 14-year follow-up study of legally blind children. *Journal of Visual Impairment and Blindness, 85*, 365–370.
- Geddie, B. E., Bina, M. J., Miller, M. M., Bathshaw, M. L., Roizen, N. J., & Lotrecchiano, G. R. (2013). Vision and visual impairment. In G. Lotrecchiano, N. Roizen & M. Bathshaw (Eds.), *Children with disabilities, Seventh edition* (pp. 169–188). Baltimore, MD: Brookes.

- Gibson, J. J. (1979). *The ecological approach to visual perception*. Boston, MA: Houghton Mifflin
- Gibson J. J. (1966). *The senses considered as perceptual systems*. Boston, MA: Houghton Mifflin.
- Kolarik, A. J., Pardhan, S., Cirstea, S., & Moore, B. C. (2017). Auditory spatial representations of the world are compressed in blind humans. *Experimental Brain Research*, 235, 597–606.
- Levtzion–Korach, O., Tennenbaum, A., Schnitzer, R., & Ornoy, A. (2000). Early motor development of blind children. *Journal of Paediatrics and Child Health*, 36, 226–229.
- Lillard, A. S. (2001). Pretend play as Twin Earth. *Developmental Review*, 21, 1–33
- Martinsen, H. (1975). The development of passivity and occurrence of stereotyped activities in congenital blind children: A presentation of a model. Manuscript, Department of Psychology, University of Oslo, Norway.
- McAllister, R., & Gray, C. (2007). Low vision: Mobility and independence training for the early years child. *Early Child Development and Care*, 177, 839–852.
- Mole, S. E., Williams, R. E., & Goebel, H. H. (2011). *The neuronal ceroid lipofuscinoses* (Batten disease), Second edition. Oxford, UK: Oxford University Press.
- Pérez-Pereira, M., & Conti-Ramsden, G. (1999). *Language development and social interaction in blind children*. Hove, UK: Psychology Press.
- Piaget, J. (1951). *Play, dreams and imitation in childhood*. London: Heineman.
- Skaggs, S., & Hopper, C. (1996). Individuals with visual impairments: A review of psychomotor behavior. *Adapted Physical Activity Quarterly*, 13, 16–26.
- Schmuckler, M. A., & Gibson, E. J. (1989). The effect of imposed optical flow on guided locomotion in young walkers. *British Journal of Developmental Psychology*, 7, 193–206.
- Tellevik, J. M., & Elmerskog, B. (2001). *The mobility and rehabilitation programme in Uganda. A sociocultural approach to working with visually impaired persons*. Trondheim: Tambartun Resource Centre.
- Tellevik, J. M., Storlieløkken M., Martinsen H., & Elmerskog B. (1998). *Spesialisten i nærmiljøet* (The specialist in the proximal environment). Oslo: Unipub.
- Vercillo, T., Tonelli, A., & Gori, M. (2017). Intercepting a sound without vision. *PLoS one*, 12(5), e0177407.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- Wan, C. Y., Wood, A. G., Reutens, D. C., & Wilson, S. J. (2010). Early but not late-blindness leads to enhanced auditory perception. *Neuropsychologia*, 48, 344–348.
- Wiener R., W., Welsh R. L., & Blasch, B. B. (2010). *Foundations of orientation and mobility, Third edition: Volume 2*. New York: AFB Press
- Willings, C. (2017). Teaching students with visual impairments: Early intervention. <https://www.teachingvisuallyimpaired.com/early-intervention.html>
- Withagen, A., Kappers, A. M., Vervloed, M. P., Knoors, H., & Verhoeven, L. (2013). Short term memory and working memory in blind versus sighted children. *Research in Developmental Disabilities*, 34, 2161–2172.
- World Health Organization. (2012). Global data on visual impairments 2010. *Geneva: World Health Organization*.

5

Dementia in Adulthood and Childhood

*Per Kristian Haugen, Stephen von Tetzchner,
Judith D. Oxley and Bengt Elmerskog*

Dementia is a syndrome due to various diseases in the brain, usually of a chronic or progressive nature, in which there is disturbance of multiple higher cortical functions, including memory, thinking, orientation, comprehension, calculation, learning capacity, language, and judgement (World Health Organization, 2018). Thus, dementia is a general term that encompasses a loss of memory and other acquired mental abilities that are severe enough to interfere with many aspects of daily life. Together with visual impairment, dementia is a core aspect of juvenile neuronal ceroid lipofuscinosis (JNCL). This chapter discusses specific aspects of dementia, including how problems with attention, memory, and other cognitive features are present in children and young people with JNCL.

The developmental trajectories of individuals with intellectual disability and dementia differ from typical development (Shapiro & Klein, 1994). The trajectory of intellectual disability reflects steady growth but has a lower curve than typical development, and the gap widens with age. The developmental trajectory of individuals with JNCL and other forms of childhood dementia initially follows the typical course for a shorter or longer interval, and then starts to decelerate and fall behind typical development, and eventually regresses below the individual's former level (Figure 5.1). There are significant individual differences in the symptomatology and developmental course of JNCL not reflected in Figure 5.1, which illustrates overall development only. The figure also illustrates why traditional methods for evaluating educational practice may not be useful for students with childhood-onset dementia.

Elderly people with adult-onset dementia may experience perceptual problems, postural instability, tremor, slowness, rigidity and other motor problems, as well as emotional or personality changes in the form of depression, anxiety or deviant social behavior (Finkel, Burns & Choen, 2000; Podell & Torres, 2011). Dementia thus influences the main functional domains and includes cognitive,

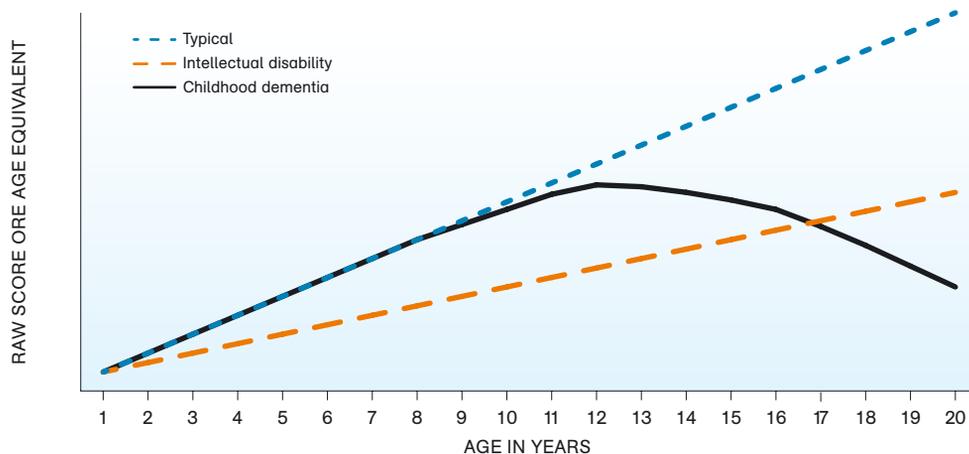


Figure 5.1 Developmental trajectories of individuals with typical development, intellectual disability and childhood dementia (after Shapiro & Klein, 1994)

The lines designate typical development, the development of children with intellectual disability and the development of children with dementia and JNCL. Development for this last group of children proceeds normally at first, then comes to a stop, and then declines.

behavioral and motor symptoms. The same features are apparent in dementia with onset in childhood (Schoenberg & Scott, 2011; Shapiro & Klein, 1994). Visual development and impairment in JNCL are discussed in Chapter 4, motor development and impairment in Chapter 7, language in Chapters 6 and 13, and emotional and behavioral problems in Chapter 27. This chapter focuses mainly on the cognitive aspects of dementia.

Dementia

Dementia is caused by physical changes in the brain, usually degeneration in the cerebral cortex, the part of the brain mainly associated with thinking, memory, action-planning, and personality. Common dementia diseases are Alzheimer disease, vascular dementia, dementia with Lewy bodies, Parkinson disease and Huntington disease (Alzheimer Europe, 2013; Engedal & Haugen, 1993; Harvey, Skelton-Robinson & Rossor, 2003). Childhood dementia is rare and less known, but several diseases imply a decline in cognitive functioning in childhood after a period of typical functioning, some with onset after a short period of normal functioning, others with onset later in childhood or adolescence (Schoenberg & Scott, 2011; Shapiro & Klein, 1994). Regardless of age, dementia represents a major challenge for the individual with the disease, and for his or her family.

Dementia is usually classified as either primary or secondary. In *primary dementia*, cognitive decline is the main symptom. Most of the primary dementia disorders, such as Alzheimer disease or frontotemporal dementia, are of a progressive nature. The symptoms emerge over time, from mild cognitive difficulties to complete dependence on assistance in all aspects of daily life. Dementia without additional symptoms is most common among older people.

In *secondary dementia*, cognitive decline is one of several symptoms and usually appears in a later phase of the disease. Secondary dementia may be related to brain tumor, Parkinson disease, acquired immune deficiency syndrome (AIDS), normal pressure hydrocephalus or subdural hematoma. Some rare neurodegenerative, autoimmune or inflammatory disorders will primarily affect people under the age of 45 (Kelley, Boeve, & Josephs, 2009). In diseases with onset in childhood, dementia is often regarded as secondary and part of a larger set of symptoms, such as in Nieman-Pick disease (Mengel et al., 2017) and mucopolysaccharidose disorder (Shapiro, Escolar, Delayney, & Mitchell, 2017). Although JNCL usually starts with symptoms of visual impairment, visual impairment is not in itself a cause of dementia. The prominent role of the cognitive decline and other features of dementia over the course of the disease suggest that dementia in JNCL might be considered primary.

A diagnosis of dementia requires that a person exhibits changes in functioning caused by a documented brain disease. There must be a decline in memory together with a reduction in one or more other cognitive functions, such as language, attention, abstraction, judgement or executive functions (Stopford, Thompson, Neary, Richardson, & Snowden, 2012). The reduction in cognitive capacity must be of such a degree that the individual is less able to manage daily life. Consciousness is not clouded and the condition must have lasted at least six months. The impairment of cognitive function is commonly accompanied, and occasionally preceded, by deterioration in emotional control, social behavior, or motivation (American Psychiatric Association, 2013; World Health Organization, 2018). The decline in memory is most evident in the learning of new information, although in more severe cases of the disease, recall of previously learned information may also be affected. In other cognitive abilities, the decline implies a deterioration in judgement and thinking, such as planning, organizing and in the general processing of information, but the person is aware of the environment (Aalten, Van Valen, Clare, Kenny, & Verhey, 2005).

It is common to describe three levels of severity. In mild dementia, the decline in cognitive abilities causes impaired performance in daily living. In moderate dementia in adulthood, the individual can perform the functions of daily living, such as shopping and handling money, but only with assistance. In severe dementia, cognitive functioning is characterized by an absence, or virtual absence, of intelligible creation of ideas (Naik & Nygaard, 2008).

In the early phase of dementia, symptoms vary considerably, depending on type of dementia. In addition, even in the same type of dementia, there may be major individual differences in symptomatology. For example, although, most persons with Alzheimer disease primarily have difficulties with memory, people with other forms of dementia may show language difficulties, apraxia, visual perception difficulties or failure in executive functions in the early phase (Koedam et al., 2010). Similar differences are apparent in childhood dementia (see below), thus emphasizing the need for careful observation of the young person with dementia to assess the overall profile of relative strengths and weaknesses (see also Chapter 10).

The following sections address three cognitive domains affected by dementia: attention, memory, and executive functioning with a view to providing basic descriptions of each, and an account of how they collectively and individually contribute to competencies and decline of competencies in education and general functioning of the young person with JNCL.

Attention

Attention refers to being aware of, focusing and processing information from the environment. In everyday life, the individual is continuously exposed to people, things and events in the environment. To understand and make sense of what is happening, the individual needs to select which stimuli are relevant and should be processed. Research suggests there are four different kinds of attention, each of which depends on a different neural network, or set of networks (Peterson & Posner, 2012; Posner, Rothbart, Sheese, & Voelker, 2014). *Focused attention* is the ability to maintain awareness over time directed at one task or event and respond specifically to one stimulus. It is believed to be mediated by the *alerting network* (Peterson & Posner, 2012). *Selective attention* refers to the ability to avoid stimuli that can distract from the focus of attention, such as cars driving by, closing of doors, or people having a conversation nearby (Lezak, Howieson, Bigler, & Trane, 2015). It is believed to be mediated by an *executive network*. *Alternating attention* requires ability to disengage and reengage one's response to stimuli in the environment (Parasuraman, 1998), and is suggested to be supported by the *orienting network*. *Divided or distributed attention* is the ability to maintain awareness of several aspects of the environment or objects at the same time. It can imply doing several things simultaneously (e.g., as when many things happen at once), or keeping track of multiple ideas simultaneously (e.g., keeping track of several elements in a verbal message) (Perry & Hodges, 1999). Distributed attention appears to rely on all three types of networks (i.e., alerting,

executive, and orienting). In summary, attention is a complex cognitive function that underlies social, educational, and daily living skills.

Problems of attention

Attention deficits are classified as a separate group of disorders (American Psychiatric Association, 2013) but problems with attention are apparent in many developmental disorders (Kerns et al., 2015). These problems are not only a matter of regulating attention better or worse than other children, but also of what children and adolescents pay attention to (Burack et al., 2016). For example, studies have found that children with autism spectrum disorders differ from their peers in terms of which aspects of a situation serve as the focus of their attention (Fan, 2013; Keehn et al., 2013). This finding suggests differences in their alerting network. Difficulties with dividing or distributing attention are often one of the first signs of dementia (Stopford et al., 2012) perhaps due to the complexity of these forms of attention. Attention and distractibility may be affected by problems related to vision (Tadi, Spring, & Dale, 2009).

Memory

Memory represents the preservation or retrieval of knowledge and of events that took place a shorter or longer time ago, often many years back in time. Memory is an essential element in all learning and necessary for the management of everyday tasks and social interaction. Memory allows the mind to use earlier experiences and makes it possible for the individual to anticipate the future and create continuity between the past, present and future. Memory problems appear in many developmental disorders (Peterson, Jones, Stephens, Gözenman, & Berryhill, 2016), and decline in memory functions is a defining feature of all forms of dementia.

The memory systems

The human memory consists of several elements with different functions. *Working memory* stores information for a very short time (seconds or minutes) and processes or organizes the information, as when searching for something or remembering a telephone number while dialing. Working memory has limited capacity. The functions of working memory are related to attention, the here-and-now and what the individual is aware of in the moment. At any given time, working memory contains what is needed in the moment. The material

held in working memory may be transferred to long-term memory or become forgotten after a few seconds. Working memory registers information from both the environment and long-term memory, and thus helps tie together new and already stored information and forms an integral part of all learning and thinking (Cowan, 2014).

Long-term memory has two parts: procedural memory and declarative memory. *Procedural memory* contains action schemas, how to perform something that is learned, including automatized actions such as bicycling, swimming or reciting the lyrics of a popular song. *Declarative memory* is divided into semantic memory and episodic memory. *Semantic memory* comprises general knowledge, facts, what the person knows, such as the names of capitals in Europe or the fact that an English mile is 1609 meters. *Episodic memory* is about the experience of the self and the quality of one's own memories. It includes such things as details from a conversation one was involved in the day before, what happened at the appointment with the doctor a week ago, or the summer vacation enjoyed many years ago. Episodic memories also have qualities such as color, shape, image, motion and sound. The content of episodic memory implies a feeling of authenticity, of "having been there", and of personal participation and involvement. The life history or *personal narrative* of the individual is made up of episodic memories (Daselar & Cabeza, 2008). Some researchers believe that semantic and episodic memory constitute two different memory systems, whereas others maintain that all the content of the declarative memory is semantic but that events experienced by the self have an added emotional quality (Parkin, 1997).

Long-term memory stores information over time, in some cases throughout life. It is considered to have unlimited capacity and constitutes the individual's personal and cultural knowledge base. However, to make use of what is stored, the individual must be able to relate the content to reality. Retrieval from memory can be accomplished through recall or recognition, such as when recognizing a voice or an object, or recalling relevant information when it is needed (Schneider, 2015). Recall is a conscious process of recreation or reconstruction, the individual tells or shows what was observed or experienced, and the focus of attention and emotional state will to some extent determine what is being recalled. By contrast, recognition can proceed unconsciously, as when something the person perceives elicits memories. However, forgetting and unlearning are also important functions of the memory system. They are necessary, enabling the child or adolescent to adapt to constantly changing environments, instead of continuing to act as if the environment remains the same (Bauer, 2014; Cuevas, Rajan, Morasch, & Bell, 2015).

Memory and dementia

In everyday life, the selection and processing of information, and the meaning-making process, may make heavy demands on working memory. Impairments in these skills, which reflect deficits in working memory, typically represent early signs of dementia. In addition, problems with episodic memory may be apparent early in dementia, and memory for recent events is often more affected than memory for personal events from the past. Semantic memory, often relatively intact in the initial phase, may nevertheless appear reduced in mild and moderate dementia. Procedural memory is usually the least affected in the early phases of dementia (Hodges, 2000).

The use of recall strategies is usually consciously executed and demands allocation of internal attentional resources, whereas the use of recognition strategies can proceed automatically, without any conscious effort, and dependent on the exploitation of external clues to retrieve memories. However, a person can actively recruit recognition strategies to help in memory retrieval (e.g., purposefully scanning shelves in supermarket for items one forgot to put on the shopping list, or on the list one forgot to bring!). Recognition strategies are preserved longer in dementia than recall strategies, and change in recognition is usually first noticeable in the moderate and severe phases. Recall is usually affected early also in mild degrees of dementia, and may decrease rapidly as the disease develops (Helkala, Laulumaa, Soininen, & Riekkinen, 1988; Van Liew et al., 2016). The name of a former classmate at primary school may be very difficult to recall, even if he or she is readily recognized in the old class photograph. It may be possible to observe for example that someone has heard a voice or seen a face, even when it is clear the person has not consciously registered that recognition has taken place.

The fact that recognition tends to be more intact than recall is of great importance when adapting the environment and encouraging people with dementia to be mentally and physically active. An environment with a limited number of familiar people, familiar surroundings, and avoidance of unnecessary relocations and changes in the environment or in the regular routines, will contribute positively to stability and security, and thus to increased activity and well-being in individuals with dementia (Kitwood, 1997; Woods, Thorgrimsen, Spector, Royan, & Orrell, 2006).

It is a common belief among relatives and professionals that people with dementia are not capable of new learning because of the extent of their memory deficits and the need for memory in learning. However, both clinical observations and research show that people with dementia may have preserved learning abilities (Bird, 1998; Bragin et al., 2009). For instance, studies have found that people with dementia benefit from learning programs as evidenced by improved processing

speed and executive functions related to planning and organization (Kawashima et al., 2015). Moreover, there is considerable individual variation among people with dementia, with a continuum of decline from relatively intact learning capacity to increasingly reduced capacity in the moderate and severe phases of dementia. Even people with quite severe dementia who move to a nursing home will often learn to find their way around and to differentiate between the care staff they meet daily and modulate conversation and interaction in ways that reflect some limited capacity to adjust to new conditions (Müller & Mok, 2018).

Executive functions

Executive functions constitute a foundation for the individual's learning, planning and performance of everyday tasks at home, in school and at work (Blair, 2016; Carlson, Zelazo, & Faja, 2013). These functions consist of several elements, including attention and working memory. They start to develop in preschool age and continue developing throughout childhood and adolescence, and some functions even into adulthood (Best, Miller, & Jones, 2009). Since executive functions are not automatized, they require more of the individual's cognitive resources than actions that can be carried out on "automatic pilot" (Diamond, 2013). It is common to distinguish between "hot" and "cold" functions. Cold executive functions include cognitive processes, while hot executive functions involve emotional and motivational factors. It is difficult for an individual to plan and make appropriate decisions when motivation and emotional involvement are too high, but also when they are too low.

Executive functions and dementia

Decline in executive functions affects planning and organization of everyday tasks, and this is apparent in the early phase of dementia, maybe due to the reduced memory and attentional capacity, which affects the ability to reason, plan and perform actions and solve practical tasks in daily life (Kirova, Bays, & Lagalwar, 2015). In mild dementia among adults, the first signs of executive difficulties may be in handling money, washing clothes or making hot food. In moderate and severe dementia, there are increasing problems with carrying out personal activities in daily life such as getting dressed and undressed, washing oneself and eating (Giebel, Sutcliffe, & Challis, 2015).

The decline in executive functions makes it difficult for the individual to get an overview of the situation he or she is in, to distinguish between relevant and irrelevant information, and to understand complex or unexpected situations.

Decline in executive functions may also imply reduced ability to suppress and control impulses, change perspective, and make sound judgements. It affects the individual's ability to learn new skills and establish an independent life.

Dementia in JNCL

Visual impairment is usually the first noticeable sign of JNCL, although there is also evidence that a decline in cognition and language may sometimes be apparent before the visual problems have become notable. The relationship between the two is complex, and cognitive decline is usually slow during childhood, thus making it difficult to pinpoint which comes first, and how the two are related (Laabs, 1988; Santavouri et al., 1988). So, although loss of vision is typically the first to be noticed, it gradually becomes clear that the disease is not restricted to vision. A lag time of approximately two years often separates the onset of visual impairment and the onset of cognitive decline, but in some cases decline proceeds in parallel (see below). There appears to be a relationship between visual and cognitive decline, in that both cognitive abilities and vision influence the ability to recognize places and orientate in the surroundings (Cameron, 1941).

Prior research shows that children with JNCL usually show typical cognitive development and participate in age-appropriate activities in kindergarten and the first school years, except for physical activities that require adequate vision (e.g., football or running). Lamminranta and associates (2001) found that from age six to ten years, the average verbal IQ on the Wechsler Intelligence Scales for Children (WISC) decreased from 88 to 72, but there were considerable individual differences. After the early school years, reports indicate that children with JNCL tend to have difficulties following lessons at school. However, the difficulties may be attributed to coping with the visual loss and therefore not perceived as a cognitive decline by people in the environment. Seizures are present in most individuals with JNCL, and EEG findings reflect slow deterioration in brain function as the illness progresses (Kirveskari, 2001). On the average, learning difficulties become apparent at the age of 8–9 years, epilepsy at 10–11 years, and speech and language problems at 12–13 years (Uvebrandt, 2006). Visual loss and speech impairment may make assessment difficult, and because cognitive assessment of children who are blind is typically based only on verbal scales, it may be difficult to distinguish cognitive abilities from language abilities.

In the present survey, some parents reported specific memory problems already when the child was 4–5 years old, but the average age when memory problems was first observed was around ten years. At 14 years, nearly 90 percent of the adolescents with JNCL had memory problems (Figure 5.2). Around ten years, they

also showed problems learning and remembering new things (Table 5.1). However, the parents reported an average age of just under eight years for problems with following mainstream education, suggesting that general cognitive problems were noticeable earlier than the specific memory problems. Functioning that is indicative of working memory problems is often apparent at an early phase of dementia (Kessels, Overbeek, & Bouman, 2015), and it is likely that it was problems with working memory that were affecting learning and school performance. However, the contribution of visual impairment to problems in following mainstream education should also be considered. Young students with JNCL must learn new behaviors to cope with their new situation (see Chapter 16), and they are challenged to do so at the same time as learning new skills is becoming more difficult. Moreover, mainstream educators usually have limited experience with teaching students who are blind. Also given the relevance of visual information to attention, particularly in young children, any decline in attention ability will add to the list of factors that challenge learning. Taken together, this situation helps explain how multiple variables compound to amplify the decline in function observed. Some of the low sub scores observed in intelligence test performance (IQ) will be due to lack of new learning (especially in the early phases), while other scores might reflect a loss of earlier acquired knowledge, or perhaps loss of ability to allocate appropriate attention in the absence of vision. This explanation is in line with other researchers who have reported that children with JNCL develop deficits in working memory which limit their ability to learn new things (van Delden, 2009). Working memory is assumed to be important for the acquisition of language and of reading

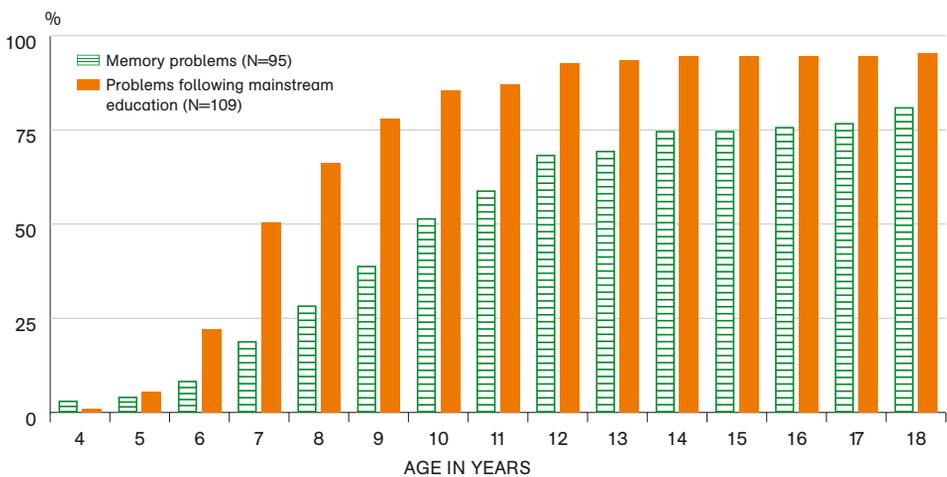


Figure 5.2 Percentage of individuals at each age from 4 to 18 years who are reported to (a) have problems with following a mainstream curriculum and (b) have problems with memory

Table 5.1 Mean age, standard deviation and age range when problems became noticeable according to parents, among individuals who have shown such problems

	Mean age	SD	Range
Problems with memory	10.5	3.9	4–23
Problems learning and remembering new things	10.7	4.1	4–27
Cognitive decline compared to peers	9.1	3.0	4–25
Problems recalling planned events of no personal significance	13.0	5.6	5–30
Problems remembering planned events of great personal significance	16.6	4.2	6–24
Problems establishing interest in new things	14.4	4.9	6–28
Problems remembering what just happened	16.0	5.5	5–28
Problems remembering daily routines	15.3	4.8	7–30
Problems remembering names of important persons	14.9	5.5	6–27
Problems remembering and maintaining strong interests	17.2	5.2	6–27
Problems maintaining previous knowledge and memories	11.7	4.4	4–25

and writing (Baddeley, 2003; Montgomery, Magimairaj, & O'Malley, 2008), and reduced working memory capacity has been suggested to make reading acquisition difficult for students with JNCL (Lou & Kristensen, 1973; but see Chapter 14). Studies have reported that on average, children with JNCL scored lower on a subtest measuring digit span than on other subscales of the WISC-R (Adams et al., 2007; Lamminranta et al., 2001). This interpretation is further supported by the fact that in the present study, cognitive decline compared with the development of peers was noted around the age of nine years.

In the present survey, however, there was considerable variation in the parents' reported age for emergence of problems with cognition and memory. The standard deviation for first problems with memory was nearly four years, which – assuming the age distribution followed the normal or Gaussian curve – means that around two-thirds of the group showed the first memory problems between age 6½ and 14½ years, and the lowest age was four and the highest 23 years. Table 5.1 shows that memory for events that had a strong significance for the individual (i.e., possibly high episodic content) on average was maintained longer (16.6 years) than memory for less important events (13.0 years). Similarly, problems maintaining previous knowledge and memories was on average observed at 11.7 years, while problems remembering and maintaining strong interests were observed at an average age of 17.2 years. This emphasizes the importance of finding educational material that is interesting and promotes participation in activities that the young person with JNCL finds engaging. Young people with JNCL may

to some extent enjoy repetition, but activities that are not intrinsically engaging may lead to lower activation of memory and less participation. Engagement thus has a dual function: increasing interest and mental activity while simultaneously preventing inattention and passivity.

Table 5.2 shows that the intercorrelations between different measures of memory were found to be in the moderate-to-high range ($r = .43 - .96$), suggesting that the individual dementia paths followed similar tracks, even if the decline started at somewhat different ages. The parents were asked to indicate the social impact of memory problems on a 5-point scale from no negative effect (1) to very high negative effect (5). An average score across ages of 3.1 ($SD = 1.1$) and a moderate correlation ($r = .37, p < .01$) suggest that the impact increased only slightly with age and the increasing degree of severity.

Last, accounting for the cognitive difficulties of dementia is crucial for the children's educational situation and needs. Dementia together with reduced vision can significantly weaken the possibility to understand and control the surroundings. Difficulties navigating the environment due to memory problems become more severe when the individual cannot make use of visual cues (Watkinson et al., 2014). However, there was no evidence prior to the present project indicating that

Table 5.2 Pearson correlations between age when different signs of memory problems first appeared (all correlations are significant, $p < .01$)

	1	2	3	4	5	6	7	8	9	10	11
1		.58	.72	.76	.54	.48	.58	.77	.43	.69	.57
2	.58		.63	.68	.58	.61	.62	.61	.56	.63	.48
3	.72	.63		.87	.75	.72	.63	.75	.58	.70	.71
4	.76	.68	.87		.74	.66	.70	.78	.56	.69	.64
5	.54	.58	.75	.74		.90	.82	.84	.81	.89	.84
6	.48	.61	.72	.66	.90		.86	.88	.69	.83	.74
7	.58	.62	.63	.70	.82	.86		.81	.70	.81	.84
8	.77	.61	.75	.78	.84	.88	.81		.69	.85	.86
9	.43	.56	.58	.56	.82	.69	.70	.69		.87	.84
10	.69	.63	.70	.69	.89	.83	.81	.85	.87		.96
11	.57	.48	.71	.64	.84	.74	.84	.86	.84	.96	

1) First memory problems, 2) Cognitive decline compared with peers, 3) Problems maintaining earlier knowledge and memories, 4) Problems learning and remembering new things, 5) Problems remembering names of important persons, 6) Problems remembering strong interests, 7) Problems establishing new interests, 8) Problems planned recalling events of no personal significance, 9) Problems recalling planned events of great personal significance, 10) Problems remembering what just happened, 11) Problems remembering daily routines

memory problems had been explicitly considered together with visual impairment by educational researchers, educational authorities, schools or JNCL counselors. Visual impairment and cognitive decline seem to have been approached as two separate phenomena. When adaptation does not consider the interaction effect of the visual impairment and cognitive decline, the result may be non-optimal physical and social environmental adaptations.

Dementia and practice related to JNCL

The main consequence of dementia is reduced cognitive capacity, and research on dementia has been concerned mainly with deficits and decay. Less attention has been given to the two issues of how people with dementia experience and evaluate their own situation, and how people in the environment can support and contribute to maintenance of intact resources and abilities. Although people with dementia, by definition have a brain disease, they may still respond actively and adapt to the disease (Cheston & Bender, 1999). Research on dementia is thus not about persons who passively develop symptoms but rather about persons who try to cope with a gradually changing life situation and control their life as the disease progresses (Beattie et al., 2004; Johannessen & Möller, 2013).

The aim of this chapter is to provide information to support families and professionals. A clear knowledge of the typical onset and course of dementia in individuals with JNCL, as well as the variation that exists, can help in several important ways. First, it can provide parents and professionals with information that can identify the help and support that a child or young person with JNCL may need; second, it emphasizes not only that help is needed, but also that help can be utilized; last, it can contribute to create awareness of the individual's *zone of developmental maintenance* (see Chapter 2), within which support can accomplish the most. The concept of dementia, viewed as creating this zone of developmental maintenance, is a foundation throughout this book in seeking to understand and support individuals with JNCL and in discussing educational tools and strategies, as well as social relations and emotional reactions. To preserve identity, self-esteem and function from day to day, persons with JNCL and other types of dementia will try to maintain a balance between the desire to maintain status quo and to adapt to the changes that occur. The challenge for parents and professionals is to see the possibilities and opportunities, even when many cognitive functions are in the process of being reduced or have been lost. People with dementia become increasingly dependent on adaptation of the environment to meet their needs. It is important that parents and professionals who help and support individuals with dementia are attentive to the pace of the decline and to what kind of help the

individual wants and needs, how he or she wants the help and when they want it (Clare, 2005). The ultimate aim will be for professionals and parents to promote maximum development and maintain use of abilities and resources throughout the course of the disease despite its constraints.

The changes in the brain of individuals with JNCL that lead to dementia constrain the possibilities for new learning and management of daily activities. As the disease progresses and the changes become more evident, the person's need for help and care increases. However, a singular focus on the brain disease may lead to overlooking possibilities that can promote growth and development, as well as possibilities for maintaining abilities and skills. There are powerful positive effects accruing to participating in activities that are of personal interest and to active engagement in motivating activities. Persons with dementia have residual abilities that should be respected: they can convey how they feel, they can share their own insights, they have resources to master, and they can benefit from other people listening, supporting and giving advice. In summary, they have important residual capabilities.

A JNCL diagnosis implies many challenges for the individual and the family. The focus will inevitably change from acquiring new competence to maintaining already established skills as the disease progresses (von Tetzchner, Fosse, & Elmerskog, 2013). It is a difficult but essential task to create an environment that is both stimulating and secure for individuals who have a reduced capacity not only for new learning, but also for coping with the new challenges posed by the ordinary daily activities (e.g., secondary to changes in functional vision). Providing appropriate support requires insight and interest in the individual's situation. Persons with dementia rely not only on receiving knowledge and help from others, but also on the helpers' commitment and engagement. Those who are responsible for providing support (e.g., organization and adaption of measures) are likely to find these responsibilities demanding, but at the same time both challenging and meaningful.

Childhood dementia constitutes a theoretical and practical framework for educational interventions where it is possible to consider windows of opportunity to meet the ongoing and anticipated declines and challenges. It is all about the foundation for providing interventions to promote the best possible quality of life for persons with JNCL. Children, adolescents and young adults with JNCL and other types of dementia disorders are in need of an adapted curriculum compared to other persons of same age. The zone of developmental maintenance (see Chapter 2) and the principles of selection, optimization and compensation (Baltes, 1987) are among the useful tools for practitioners who are working with children and young people with JNCL. The educational strategies described in several chapters in this book (hastened learning, precautionary learning, skill-based learning, participation-based learning and life flow) are based on knowledge about the typical course and variation in JNCL, the zone of developmental maintenance, and Baltes' model.

References

- Aalten, P., Van Valen, E., Clare, L., Kenny, G., & Verhey, F. (2005). Awareness in dementia: a review of clinical correlates. *Aging and Mental Health, 9*, 414–422.
- Adams, H. R., Kwon, J., Marshall, F. J., de Blicke, E. A., Pearce, D. A., & Mink, J. W. (2007). Neuropsychological symptoms of juvenile-onset batten disease: Experiences from 2 studies. *Journal of Child Neurology, 22*, 621–627.
- Alzheimer Europe (2013). *Dementia in Europe Yearbook 2013*. Downloaded from <https://www.alzheimer-europe.org/Publications/Dementia-in-Europe-Yearbooks>.
- American Psychiatric Association (2013). *Diagnostic and Statistical Manual of Mental Disorders, Fifth edition (DSM-5)*. Washington, DC: American Psychiatric Association.
- Baddeley, A. D. (2003). Working memory and language: A review. *Journal of Communication Disorders, 36*, 189–208.
- Baltes, P. B. (1997). On the incomplete architecture of human ontogeny: Selection, optimization, and compensation as foundation of developmental theory. *American Psychologist, 52*, 366–380.
- Bauer, P. J. (2014). The development of forgetting: Childhood amnesia. In P. Bauer & R. Fivush (Eds.), *The Wiley handbook on the development of children's memory, Volume I-II* (pp. 513–544). London, UK: Wiley.
- Beattie, A., Daker-White, G., Gilliard, J., & Means, R. (2004). 'How can they tell?' A qualitative study of the views of younger people about their dementia and dementia care services. *Health and Social Care in the Community, 12*, 359–368.
- Best, J. R., Miller, P. H., & Jones, L. L. (2009). Executive functions after age 5: Changes and correlates. *Developmental Review, 29*, 180–200.
- Bird, M. (1998). Clinical use of preserved learning capacity in dementia. *Australasian Journal on Ageing, 17*, 161–166.
- Blair, C. (2016). Executive function and early childhood education. *Current Opinion in Behavioral Sciences, 10*, 102–107.
- Bragin, V., Chemodanova, M., Vaysman, A., Bragin, I., Chernyavskyy, P., Grinayt, E., & Aliev, G. (2009). Preservation of learning abilities in people with dementia and depression with different level of cognitive impairment. *Alzheimer's and Dementia: The Journal of the Alzheimer's Association, 5*, P322.
- Burack, J. A., Russo, N., Kovshoff, H., Palma Fernandes, T., Ringo, J., Landry, O., & Iarocci, G. (2016). How I attend – not how well do I attend: Rethinking developmental frameworks of attention and cognition in autism spectrum disorder and typical development. *Journal of Cognition and Development, 17*, 553–567.
- Cameron, D. E. (1941). Studies in senile nocturnal delirium. *Psychiatric Quarterly, 15*, 47–53.
- Carlson, S. M., Zelazo, P. D., & Faja, S. (2013). Executive function. In P. Zelazo (Ed.), *The Oxford handbook of developmental psychology, Volume 1* (pp. 706–743). Oxford, UK: Oxford University Press.
- Cheston, R., & Bender, M. (1999). *Understanding dementia: The man with the worried eyes*. Philadelphia: Jessica Kingsley.
- Clare, L., Roth, I., & Pratt, R. (2005). Perceptions of change over time in early-stage Alzheimer's disease: Implications for understanding awareness and coping style. *Dementia, 4*, 487–520.

- Cowan, N. (2014). Short-term and working memory in childhood. In P. J. Bauer & R. Fivush (Eds.), *The Wiley handbook on the development of children's memory, Volume 2* (pp. 202–229). Chichester, UK: Wiley.
- Cuevas, K., Rajan, V., Morasch, K. C., & Bell, M. A. (2015). Episodic memory and future thinking during early childhood: Linking the past and future. *Developmental Psychobiology*, *57*, 552–565.
- Daselaar, S. M., & Cabeza, R. (2008). Episodic memory decline and healthy aging. In J. H. Byrne & H. Eichenbaum (Eds.), *Learning and memory: A comprehensive reference* (pp. 577–599). Oxford, UK: Elsevier.
- Diamond, A. (2013). Executive functions. *Annual Review of Psychology*, *64*, 135–168.
- Engedal, K., & Haugen, P. K. (1993). The prevalence of dementia in elderly Norwegians. *International Journal of Psychiatry*, *8*, 565–570.
- Fan J. (2013) Attentional network deficits in autism spectrum disorders. In J. D. Buxbaum & P. R. Hof (Eds.), *The neuroscience of autism spectrum disorders* (pp. 281–288). Oxford, UK: Elsevier.
- Finkel, S. I., Burns, A., & Cohen, G. (2000). Behavioral and psychological symptoms of dementia (BPSD): A clinical and research update – Overview. *International Psychogeriatrics*, *12*, 13–18.
- Giebel, C. M., Sutcliffe, C., & Challis, D. (2015). Activities of daily living and quality of life across different stages of dementia: A UK study. *Aging and Mental Health*, *19*, 63–71.
- Harvey, R. J., Skelton-Robinson, M., & Rossor M. N. (2003). The prevalence and causes of dementia in people under the age of 65 years. *Journal of Neurology, Neurosurgery, and Psychiatry*, *74*, 1206–1209.
- Helkala, E. L., Laulumaa, V., Soininen, H., & Riekkinen, P. J. (1988). Recall and recognition memory in patients with Alzheimer's and Parkinson's diseases. *Annals of Neurology*, *24*, 214–217.
- Hodges, J. R. (2000). Memory in the dementias. In E. Tulving & F. I. M. Craik (Eds.), *The Oxford handbook of memory* (pp. 441–459). New York, NY: Oxford University Press.
- Howe, M. L. (2015). Memory development. In R. M. Lerner, L. S. Liben and U. Müller (Eds.), *Handbook of child psychology and developmental science, Seventh edition, Volume 2: Cognitive processes* (pp. 203–249). Hoboken, NJ: Wiley.
- Johannessen, A., & Möller, A. (2013). Experiences of persons with early-onset dementia in everyday life: A qualitative study. *Dementia*, *12*, 410–424.
- Kawashima, R., Hiller, D. L., Sereda, S. L., Antonczak, M., Serger, K., Gannon, D., et al. (2015). SAIDO learning as a cognitive intervention for dementia care: A preliminary study. *Journal of the American Medical Directors Association*, *16*, 56–62.
- Keehn, B., Müller, R. A., & Townsend, J. (2013). Atypical attentional networks and the emergence of autism. *Neuroscience and Biobehavioral Reviews*, *37*, 164–183.
- Kelley, B. J., Boeve, B. F., & Josephs, K. A. (2009). Cognitive and noncognitive neurological features of young-onset dementia. *Dementia and Geriatric Cognitive Disorders*, *27*, 564–571.
- Kerns, C. M., Kendall, P. C., Zickgraf, H., Franklin, M. E., Miller, J., & Herrington, J. (2015). Not to be overshadowed or overlooked: Functional impairments associated with comorbid anxiety disorders in youth with ASD. *Behavior Therapy*, *46*, 29–39.
- Kessels, R. P. C., Overbeek, A., & Bouman, Z. (2015). Assessment of verbal and visuospatial working memory in mild cognitive impairment and Alzheimer's dementia. *Dementia and Neuropsychologia*, *3*, 301–305.
- Kirova, A. M., Bays, R. B., & Lagalwar, S. (2015). Working memory and executive function decline across normal aging, mild cognitive impairment, and Alzheimer's disease. *BioMed Research International*, *2015*, 748212.

- Kirveskari, E. (2000). Circadian rhythms and sleep in neuronal ceroid lipofuscinosis. Academic Dissertation, University of Helsinki.
- Kitwood, T. (1997). *Dementia reconsidered: The person first*. Buckingham, UK: Open University Press.
- Koedam, E. L. G. E., Lauffer, V., van der Vlies, A. E., van der Flier, W. M., Scheltens, P., & Pijnenburg, Y. A. L. (2010). Early-versus late-onset Alzheimer's disease: More than age alone. *Journal of Alzheimer's Disease*, *19*, 1401–1408.
- Laabs, R. (1988). Zur klinischen Variabilität der Juvenilen Neuronalen Ceroid-Lipofuscinose. Versuch einer quantitativen Beschreibung des Verlaufs bei 17 Patienten (On the clinical variation in juvenile neuronal ceroid lipofuscinosis. An attempt to quantitative description of the course of 17 patients). Doctoral thesis, University Hospital, Eppendorf, Germany.
- Lamminranta, S., Åberg, L.E., Autti, T., Moren, R., Laine, T., Kaukoranta, J., et al. (2001). Neuropsychological test battery in the follow-up of patients with juvenile neuronal ceroid lipofuscinosis. *Journal of Intellectual Disability Research*, *45*, 8–17.
- Lezak, M. D., Howieson D. B., Bigler E. D., & Trane D. (2015). *Neuropsychological assessment*. Oxford, UK: Oxford University Press.
- Lou, H. C., & Kristensen, K. (1973). A clinical and psychological investigation into juvenile amaurotic idiocy in Denmark. *Developmental Medicine and Child Neurology*, *15*, 313–323.
- Mengel, E., Pineda, M., Hendriks, C. J., Walterfang, M., Torres, J. V., & Kolb, S. A. (2017). Differences in Niemann-Pick disease Type C symptomatology observed in patients of different ages. *Molecular Genetics and Metabolism*, *120*, 180–189.
- Montgomery, J. W., Magimairaj, B., & O'Malley, M. (2008). The role of working memory in typically developing children's complex sentence comprehension. *Journal of Psycholinguistic Research*, *37*, 331–354.
- Naik, M., & Nygaard, H. A. (2008). Diagnosing dementia – ICD-10 not so bad after all: A comparison between dementia criteria according to DSM-IV and ICD-10. *International Journal of Geriatric Psychiatry*, *23*, 279–282.
- Parkin, A. J. (1997) The development of procedural and declarative memory. In N. Cowan (Ed.), *The development of memory in childhood* (pp. 113–137). London: Psychology Press.
- Perry, R. J., & Hodges, J. R. (1999). Attention and executive deficits in Alzheimer's disease: A critical review, *Brain*, *122*, 383–404.
- Peterson, S. E., & Posner, M. I. (2012). The attention system of the human brain: 20 years after. *Annual Review of Neuroscience*, *35*, 75–89.
- Peterson, D. J., Jones, K. T., Stephens, J. A., Gözenman, F., & Berryhill, M. E. (2016a). Childhood memory: An update from the cognitive neuroscience perspective. In W. T. O'Donohue & M. Fanetti (Eds.), *Forensic interviews regarding child sexual abuse* (pp. 81–105). Cham, Switzerland: Springer.
- Peterson, S. E., & Posner, M. I. (2012). The attention system of the human brain: 20 years after. *Annual Review of Neuroscience*, *35*, 75–89.
- Podell, K., & Torres, K. (2011). Affective symptoms in early-onset dementia. *Neurologic Clinics*, *29*, 99–114.
- Posner, M. I., Rothbart, M. K., Sheese, B. S., & Voelker, P. (2014). Developing attention: Behavioral and brain mechanisms. *Advances in Neuroscience*, *2014*, 405094.
- Pozuelos, J. P., Paz-Alonso, P. M., Castillo, A., Fuentes, L. J., & Rueda, M. R. (2014). Development of attention networks and their interactions in childhood. *Developmental Psychology*, *50*, 2405–2415.

- Santavuori, P., Heiskala, H., Westermarck, T., Sainio, K., Moren, R., Opitz, J. M., Reynolds, J. F., & Pullarkat, R. K. (1988). Experience over 17 years with antioxidant treatment in Spielmeier-Sjögren disease. *American Journal of Medical Genetics Part A*, 31 (S5), 265–274.
- Schneider, W. (2015). *Memory development from early childhood through emerging adulthood*. Cham, Switzerland: Springer.
- Schneider, W., & Pressley, M. (1997). *Memory development between 2 and 20, Second edition*. Mahwah, NJ: Erlbaum.
- Schoenberg, M. R., & Scott, J. G. (2011). Cognitive decline in childhood or young adulthood. In M. R. Schoenberg & J. G. Scott (Eds.) *The little black book of neuropsychology: A syndrome-based approach* (pp. 839–861). New York, NY: Springer.
- Shapiro, E. G., Escolar, M. L., Delaney, K. A., & Mitchell, J. J. (2017). Assessments of neurocognitive and behavioral function in the mucopolysaccharidoses. *Molecular Genetics and Metabolism*, 122, 8–16.
- Shapiro, E. G., & Klein, K. A. (1994). Dementia in childhood: Issues in neuropsychological assessment with application to the natural history and treatment of neurodegenerative storage diseases. In M. G. Tramonata & S. R. Hooper (Eds.), *Advances in child neuropsychology, Volume 2* (pp. 119–171). New York, NY: Springer.
- Stopford, C. L., Thompson, J. C., Neary, D., Richardson, A. M., & Snowden, J. S. (2012). Working memory, attention, and executive function in Alzheimer’s disease and frontotemporal dementia. *Cortex*, 48, 429–446.
- Tadić, V., Pring, L., & Dale, N. (2009). Attentional processes in young children with congenital visual impairment. *British Journal of Developmental Psychology*, 27, 311–330.
- Uvebrant, P. (2006). The interaction between neurology and education in juvenile Batten disease. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 95–97). Örebro, Sweden: Swedish Institute for Special Needs Education.
- van Delden, G. W. (2009). Education for children with Batten disease. Presented at the International Council for the Education and rehabilitation of People with visual Impairment Europe Conference, Dublin, Ireland, July 5–10, 2009.
- Van Liew, C., Santoro, M. S., Goldstein, J., Gluhm, S., Gilbert, P. E., & Corey-Bloom, J. (2016). Evaluating recall and recognition memory using the Montreal Cognitive Assessment: Applicability for Alzheimer’s and Huntington’s diseases. *American Journal of Alzheimer’s Disorders and Other Dementias*, 31, 658–663.
- Vogel, A., Stokholm, J., Gade, A., Andersen, B. B., Hejl, A. M., & Waldemar, G. (2004). Awareness of deficits in mild cognitive impairment and Alzheimer’s disease: Do MCI patients have impaired insight? *Dementia and Geriatric Cognitive Disorders*, 17, 181–187.
- von Tetzchner, S., Fosse, P., & Elmerskog, B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta – Molecular Basis of Disease*, 1832, 1894–1905.
- Watkinson, S. (2014). *Older people with visual impairment – Clinical management and care*. Manchester, UK: M&K Publishing.
- Woods, B., Thorgrimsen, L., Spector, A., Royan, L., & Orrell, M. (2006). Improved quality of life and cognitive stimulation therapy in dementia. *Aging and Mental Health*, 10, 219–226.
- World Health Organization (2018). *International classification of diseases, Eleventh edition*. Available from <http://www.who.int/classifications/icd/en/>

6

Language Development and Attrition in Juvenile Neuronal Ceroid Lipofuscinosis

Anne-Grethe Tøssebro and Stephen von Tetzchner

Language development may be defined as the process by which children come to comprehend and use linguistic forms and structures through interactions with more competent children and adults in a variety of social and cultural circumstances (von Tetzchner, 2019). Language distinguishes humans from other species, and is the basis for the child's entry into society. Human language consists of linguistic symbols that represent social conventions to draw attention to specific people, things, events and ideas, and a grammar, which allows the symbols to be arranged in patterns according to certain conventions – sentence structures – that create meaning beyond the individual symbols themselves (Tomasello, 2006). Language makes advanced communication possible about a range of topics for a variety of purposes, as well as reflection on the past and the future, creating continuity between past, present and future. Written language allows for the organization and storage of knowledge beyond the limits of each individual's memory (see Chapter 14).

Language has the same functions in children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL), but their developmental course is unique in that it typically includes both language growth and attrition, and hence implies a need for measures that can support and maintain communication (see Chapter 13).

Language comprehension and production

Language use implies both comprehension and production, and the two processes make different demands on the user. When inferring meaning from language produced by others, the child has to recognize the words' perceptual patterns (visual, auditory or tactile), the communicative intentions, and the social patterns of the situation. Language production requires a purpose or meaning and an ability to articulate, that is, mastery of the mechanics of speech or another mode

of production. Expressive language may represent an initiative to communicate, relaying opinions, emotions, interests and stories, or a response to communicative initiatives from others, such as answering questions in a dialogue. Language comprehension and production may follow the same or somewhat different developmental courses (Bloom, 1974; Bates, Bretherton, & Snyder, 1988). Language attrition may affect both comprehension and expression, initiation and responding, but in different ways, at different times, and at different speeds. In typical development, children learn to master the speech sounds and phonemic contrasts of the language, as well as the syllable shapes and prosodic patterns. This developmental period will include errors observed from the onset of first words and into school age. Attrition of speech ability might appear in the form of persisting developmental errors (such as final consonant deletion of words or difficulty with particular sounds), or complete loss of contrast between sounds (the child produces different words that sound the same to the listener; e.g., *go* and *goat* both sound like *go*). Speech attrition can also affect the fluency, resulting in stuttered speech.

Language development in children with JNCL

There is a significant body of research on language development in children with congenital blindness (e.g., Fraiberg, 1977; Perèz-Pereira & Conti-Ramsden, 1999; Pijnacker, Vervloed, & Steenbergen, 2012; Tadic, Pring, & Dale, 2010). Despite the lack of visual information, children who are blind without additional impairments show a relatively normal language development, although many words may have a somewhat different conceptual content and usage than in sighted children. For example, "look" may not refer to vision but to experience in a more general sense (Landau & Gleitman, 1985). Nonetheless, language is the main source of information about social factors and the physical world, and early language skills are one of the strengths of many children who are blind. Moreover, their social interaction is mainly based on language use, and many children who are blind are rather talkative.

There is less research on language development among children who become blind in middle childhood. The onset of visual impairment for most children with JNCL occurs between 5 and 8 years of age (see Chapter 4). Their early language development therefore proceeds in a typical way, with no signs of the upcoming language problems. They use visual cues in the same way as other sighted children, and this is reflected in memory, conceptual structure and language use. This means that almost all children with JNCL are in a normal process of language development at the time when visual impairment and emerging dementia start to affect their language development and language attrition begins.

Language attrition in JNCL

All individuals with JNCL will experience language attrition but both the age when language problems become noticeable and the nature of the problems vary considerably. In particular, it is important to distinguish between comprehension and expression, and in general, expressive language seems to be more affected and at an earlier age than language comprehension. The expressive problems make it difficult for the family, peers and others to understand what the person with JNCL is trying to communicate, and they may have to rely on contextual factors when interpreting what the person is saying. Familiar people will understand more than communication partners who do not know the person so well.

Expressive language

Table 6.1 shows the age when decline became observable for core aspects of language attrition in the present study (see Appendix A). The average age when problems with speech became noticeable was 13 years and ranged from 2 to 25 years. Similar results have been reported in other studies (Bendixen, 1996; Kohlschütter, Laabs, & Albani, 1988). Among the 91 participants in the present study who were reported to have shown speech problems, six showed the first signs of language problems at the age of five or earlier, while five were 21 years or older when these symptoms first appeared. None of the speech problems were found in 100 percent of the participants. Fifteen children and young people had not developed unclear speech at the time of the surveys and interviews. For some this may be due to young age but the results also reflect the considerable differences in age of onset for the diverse speech problems. One parent described her daughter's development in this way:

Speech slowly left her. She went from being able to speak at will, to speaking when spoken to, to short phrases, to one word response as yes or no, to complete non-verbal (...) but she can still make noises if she is agitated or uncomfortable.

The decline of speech in young people with JNCL is complex. The pattern of speech problems differed considerably among the participants in the present study, and most had more than one speech problem. Articulation problems were common, leading to reduced speech intelligibility, that is, the partners' recognition of word forms. Eighty-three participants were reported to have developed speech that was imprecise but usually intelligible to unfamiliar people at an average of 14 years. Sixty-eight participants had developed speech that was unclear and not usually

Table 6.1 Number of individuals who show core expressive language and speech problems, and average age when observed (N=111)

Observed speech (S) and expressive language (EL) problems	n	Mean age of onset if occurred	SD	Range
First sign of speech problem	91	13.0	4.7	2–25
Stuttering (S)	47	10.8	4.3	4–20
Using wrong words (EL)	36	12.6	4.8	6–26
Word finding problems (EL)	83	12.7	4.8	4–25
Mumbling (S/EL)	54	13.9	4.2	5–24
Only short sentences (EL)	56	17.2	3.9	10–25
Limited vocabulary, same words repeated (S/EL)	48	18.2	4.1	10–27
Voice weak and difficult to hear (S)	24	20.6	4.3	13–26
Problems calling attention with speech (S)	38	18.2	4.1	10–30
Only single words (EL)	42	20.6	3.9	12–30
Imprecise but usually intelligible to unfamiliar people (S)	83	14.0	4.3	3–25
Unclear and not usually intelligible to unfamiliar people out of context (EL/S)	68	16.8	3.7	7–26
Speech no longer comprehensible (S/EL)	32	20.3	3.6	14–30
No production of speech (S/EL)	23	22.5	3.6	15–28

comprehensible to unfamiliar people outside a known context at the average age of onset of 16.8 years, while the average age for having developed unintelligible speech was 20.3 years. Twenty-three participants had stopped producing speech at an average age of 22.5 years. Some participants were unable to use speech to gain attention from others by an average age of 18.2 years, and in some participants, the voice had become very weak and utterances almost inaudible by 20.6 years. The communication partners' problems with comprehending unclear speech increase in noisy conditions. However, for all the symptoms, the age range was considerable. For example, age of onset of imprecise but usually intelligible speech ranged from 3 to 25 years, and age when speech had become incomprehensible ranged from 14 to 30 years. Several parents described this process:

The pronunciation of many words become almost the same. Very often other people misunderstand.

The words simply don't come out, even if she keeps trying.

Similar observations were made by staff:

His speech is very slurred. People who do not know him do not understand him. He repeats single important words until the counterpart guesses the correct word.

Together with imprecise but intelligible speech, problems with word finding were most frequent. Eighty-three participants showed this symptom at an average age of 12.7 years, which is a common feature in developmental language disorders (Best, 2005; Messer & Dockrell, 2006) and dementia (Bang, Spina, & Miller, 2015; Klima & Kuca, 2016; Rohrer et al., 2008). Use of wrong words was observed around the same age but this was much less frequent (36 participants). There are similar reports of idiosyncratic use of words without showing awareness of it, and odd constructions like «confirmation ingredients» and «world champion dancing dress» (Chaffey, 1987; Laabs, 1988; von Tetzchner, 2006). Idiosyncratic use of words make the person's expressive language difficult to understand.

Dysfluency (cluttering or stuttering) is considered a typical feature of language development in children and young people with JNCL (Gayton, 1982; von Tetzchner, 2006) and was observed early (average age 10.8 years) but only for 47 participants. Mumbling was also a relatively early feature (average age 13.9 years) for 57 participants, about the same proportion as for dysfluency. The intelligible expressive vocabulary may decrease and repetitions of the same words had become prominent by an average age of 18.2 years (table 6.1), as described by one parent:

Repetitious (has to start again many times). Often he is stuck with the first word.

Information from bereaved families, which covers the whole life span of 33 participants with JNCL, indicates that stuttering never occurred in 16 of these participants. The results indicate that speech dysfluency is quite common in young people with JNCL but also that about half of the people with the disease never develop dysfluency, although they have other speech problems. Similarly, only 19 (57%) of the bereaved parents told about complete loss of intelligible speech in their child at some point in time. This indicates that a considerable proportion of adolescents and adults with JNCL retain some intelligible speech throughout life, and that they at least to some degree can make themselves understood by their family and others who know them well.

Problems with sentence construction were observed in many participants, typically with onset in late adolescence. About half of the participants used only short sentences, observed by an average age of 17.2 years and about one third had begun to use mainly single-word utterances, observed by the age of 20.6 years.

There are very few detailed descriptions of the expressive language development and attrition in children and young people with JNCL. To our knowledge, there is only one linguistic study. Chaffey (1987) analyses the expressive language of her daughter Christine. The first signs of language attrition were problems with finding words, especially nouns. Around age 16 years, she started to stutter and often repeated what the other said, maybe reflecting problems with comprehension or word finding. Sentence structure was mainly maintained to 18 years, when Christine started to make errors in negation. She also began to make errors in sentences with complex syntax, simplified the sentences and used fewer syntactic and more analytic constructions. Her utterances involving negation comprised both syntactic and semantic relations, but they were arranged in unusual manners. When making a sentence, she seemed to start with a positive sentence (which initially is simpler) and then add a simple negation, such as *All animals with fur she can stand not* instead of «she cannot stand any animals with fur» (allergic) or *Always I want not you leave* instead of «I never want you to leave» (translated from Norwegian). Many of Christine's sentence constructions may also be found in the language of younger children, and are also typical of language attrition in general (Chaffey, 1987). Even when each word is intelligible, unusual syntactic constructions may make the meaning of the person's utterances difficult to understand.

The decline in expressive language constitutes a major problem when persons with JNCL try to convey meaningful contributions to the dialogue. Many children with JNCL are rather talkative, and may experience comfort from conversations about past experiences (van Delden, 2009). However, even when they speak a lot, partner comprehension may be limited:

Speaks very much, knows and uses a lot of words, but is no longer understandable.

Children and young people with JNCL are not always aware of their own speech and expressive language problems and may struggle to understand why conversation partners do not give instant answers to their conversational initiatives, as described by parents and staff:

For a long time she thought that her speech was clear, that the others didn't hear well. It took a long time before she became aware that her speech wasn't that easy to understand.

She was not conscious of her speech problems.

The huge variety found in the present and other studies of expressive language in children and adults with JNCL emphasizes the need to adapt support and intervention to individual abilities and challenges in both early and later phases of the disease.

Language comprehension and expression

Reduced language comprehension is a core element of dementia (see Chapter 5), and language comprehension will decline also in children and adults with JNCL. Childhood dementia may particularly affect comprehension of abstract concepts and complex procedures. However, when topics are concrete and related to familiar everyday issues and the person's own experiences and personal interests, information from parents and professionals suggest that comprehension may be preserved even when intelligible speech is lost:

She understood language well at the age of 22 even though she could hardly express herself.

We always felt that she continued to understand a great deal even though she was unable to speak back to us.

Although language comprehension is affected by the emerging dementia, it is much better retained than speech production. In fact, comprehension of vocabulary may continue to increase even after cognitive functions have started to decline. Hearing acuity is usually not affected by the disease, but comments in the present study indicate that some young people with JNCL experience problems in processing auditory information or might become hypersensitive to sounds and find noisy environments disturbing and unpleasant. Still, hearing and language comprehension are their most important resources for understanding their environment and participating in it.

In the present study (Appendix A), parents were asked to compare their child's language comprehension in relation to expression both at the time of the survey and retrospectively from age seven years. The survey included factors that relate to expressive language, speech, or skills that could reflect a combination of these competencies. Figure 6.1 shows that comprehension and expression initially were considered equal and that comprehension gradually became relatively better than expression with age. At the age of 7 years, comprehension and expression were considered equal in 82.5 percent of the participants. By the age of 22, only 6.7 percent of the participants were considered having equal comprehension and expression of spoken language. However, it may be noted that a small group

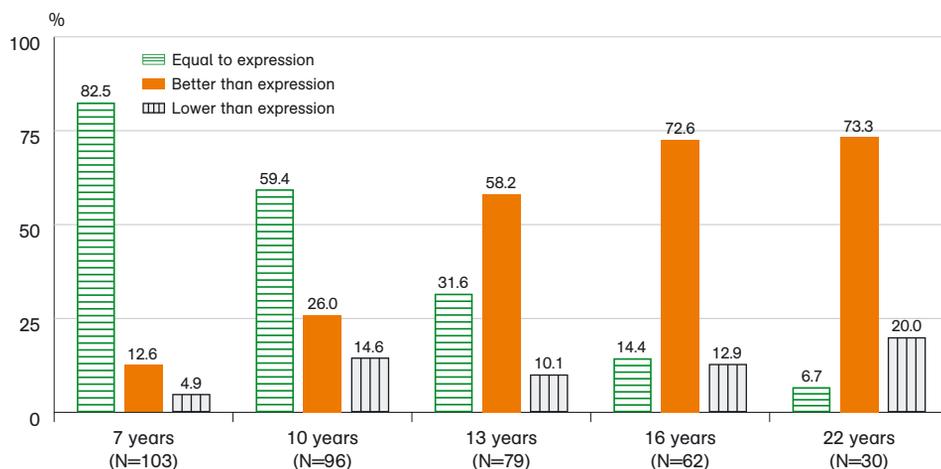


Figure 6.1 Comprehension of speech in relation to expression (percent of responses)

seemed to retain an ability to speak but had problems with comprehension and probably also expressive content.

In the questionnaires and interviews, parents and professionals described comprehension as better than the expressive ability.

Language comprehension was not his major problem. Language production was getting difficult when he was getting older.

She listened to audio books and her comprehension was good until around age 14–15. Then she started to regress, preferring younger books and more repetition.

The results thus support observations from clinical experience that the decline in speech production and expressive content is more pervasive than comprehension of spoken language (Elmerskog & Fosse, 2012; Gombault, 2010).

Both language comprehension and production are influenced by familiarity with the situation. Persons with severe visual impairment depend on non-visual contextual cues, such as what other people say and sounds from ongoing activities in the environment but will miss cues that require vision. The cognitive decline may lead to fragmented experiences, which in turn make language comprehension fragmented and lacking in coherence (von Tetzchner, 2006). For young people with JNCL, being in a familiar environment will enhance comprehension and facilitate communication. As the disease progresses, their comprehension becomes increasingly dependent on being in a situation that is familiar and does not contain

too many new elements or the kinds of noise that the individual finds distressing because of susceptibility to sensory overload. An adapted school environment that is stable and predictable is therefore important also for communication. In adulthood, the same applies to activity centers and work places.

The gap between comprehension and expressive spoken language communication also suggests that young people with JNCL may benefit from augmentative and alternative communication (AAC) intervention (see Chapter 13).

Observations and assessments

Both formal and informal assessment of individuals with severe visual impairment and dementia may be challenging and require adapted material and situations (see Chapter 10).

Language is important for all aspects of communication and daily life. When language is impaired, the promotion of communication and language is always an important target in the curriculum. Assessment of comprehension and use of language are therefore important elements of educational practice. There are various tests and checklists for evaluating language competence, both comprehension and production (e.g., Goldman & Fristoe, 2015). Many of the tests are based on picture recognition or naming and therefore are not suited for children and young people with visual impairment (Eirin & Koenig, 1997). When children's vision precludes use of such tests, alternate assessments can be obtained through use of inventories of spoken words drawn from a sample of speech in which the child is actively engaged in activities that include known words (toys, actions, people, etc.), and from parent diaries of spoken word attempts of known words (Bauman-Waengler, 2016; Velleman, 2016). For example, use of scripted activities helps assure a known context for children with highly unintelligible speech (Lund & Duchan, 1993).

Receptive language is difficult to evaluate and assess (Bishop, 2006). Tadic and associates (2010) used different tests of expressive and receptive language (e.g., CELF-3, CCC-2) with a group of children with congenital blindness, and found that a few subtests could be used for children with no vision, but also that these test results would have reduced validity.

Receptive language skills may to some extent be inferred from responses to questions or how instructions are followed, but these are typically based on spoken answers, visual discrimination or motor performance. In such observations, it may be difficult to distinguish between receptive and expressive language competence, as well as motor performance (Graven, 2018).

Expressive language is also difficult to assess formally in individuals with JNCL, but speech and problems related to expressive language are easier to observe and describe than comprehension. The different aspects of language decline in JNCL follow a recognizable course, although there are individual differences concerning time and speed of symptom occurrence. An alternative to formal assessments is to observe a young person with JNCL in a range of settings and record their language use and their responses during observations. This can be done for example annually or semiannually with standard procedures in situations that are familiar to the person.

Observations of receptive language require interpretation and analysis. It is necessary to record all attempts to elicit understanding or response, as well as words and other communicative expressions of the individual. It may be useful to observe interaction with several people, both familiar and unfamiliar partners, as the quality of the interactions may reflect the extent of the partner's personal knowledge and insight into the communicative means of the person. Registration and discussion of all cues and expressions are important also to avoid attributing intentions and attitudes that the person with JNCL does not have (Grove, Bunning, Porter, & Olsson, 1999).

Educational Development Observation (EDO)

EDO is an observation checklist for assessing educational needs and possibilities developed for use with children and young people with JNCL. Table 6.2 shows the language and communication part (see Chapter 10 for more information about the EDO). Use of the EDO for evaluation of people with JNCL should include input from a professional with comprehensive knowledge about both JNCL and EDO. In communication and other domains it is an aim to start intervention before symptoms develop, because this may allow the child to develop skills that he or she will need but not will be able to develop in later phases of the disease. The principles of hastened and precautionary learning are important when learning for the future (see Chapter 12). A professional with expertise in JNCL will have knowledge about future declines in communication and language and can guide parents and professionals in preparing the child for these declines in best possible way. The checklist should not be used solely for mapping impairment or symptoms. Rather, the intention is to guide the support team to identify the best possible interventions to compensate and optimize the individual's social participation and quality of life.

Table 6.2 The communication and language part of *Educational Development Observation Tool* (EDO)

<p>C1. Speech intelligibility (mark one alternative)</p> <ol style="list-style-type: none"> 1) Speech is understood by all, special attention from others is not required 2) Someone who knows the person well may not need to ask for repetition/clarification but someone who does not know the person well, may sometimes have to ask for clarification of a few words 3) Speech is unclear and not usually understandable to unfamiliar listeners out of context 4) Someone who knows the person well will understand simple, single words only (e.g., yes/no, etc.). 5) The person's speech cannot be understood by anyone
<p>C2. Speech (mark all that apply)</p> <ol style="list-style-type: none"> 1) Speech is normal (<i>note, if speech is normal, no other options should be marked</i>) 2) The person stutters 3) The person mumbles/has poor articulation 4) The person has problems finding the right words 5) The person uses wrong words when trying to express something 6) The person is only able to express short and simple sentences (3–6 words) 7) The person is only able to express single words 8) The person's voice is weak and difficult to hear 9) The person has very limited vocabulary, uses the same words again and again 10) The person has problems with calling on other people's attention by speech 11) Other, please describe:
<p>C3. Expressive communication modes: What expressive communication modes are used? (mark one alternative)</p> <ol style="list-style-type: none"> 1) Mostly oral communication 2) A combination of oral communication and signs/gestures/body language 3) Mostly communication via signs/gestures/body language 4) Mostly communication via technical/electronic aids or/and objects of reference 5) The person has no or almost no expressive communication
<p>C4. Expressive communication: When using expressive communication alternatives, is communication understandable? (mark one alternative)</p> <ol style="list-style-type: none"> 1) Irrelevant, no need for communication alternatives 2) Communication alternatives are imprecise but usually understandable even for unfamiliar recipients 3) Communication alternatives are unclear and not usually understandable to unfamiliar recipients out of context 4) Communication alternatives are unintelligible even to those who know the person well 5) No communication alternatives are used despite the need

C5. Language: Language comprehension versus language production/expressive communication (mark one alternative)

- 1) Language comprehension is the same as language production (normal). The person shows an equal correspondence between what the person is able to express (for instance by talking, signing, body language, using technical aids etc.) and the person's comprehension of what other people are expressing
- 2) Language comprehension is slightly better than language production. The person is from time to time showing that he/she understands more than she/he is able to express. The difference is evident but does not have a major impact on everyday living
- 3) Language comprehension is better than language production. The person is on a regular basis showing that he/she understands more than he/she is able to express. The difference has an impact on everyday living.
- 4) Language comprehension is evident, but there is no expressive communication. The person has no or almost no expressive communication but it is evident that the person from time to time understands what others are expressing
- 5) There is no expressive communication and it is difficult to assess if there is any language comprehension. The person has no or almost no expressive communication and there are no or almost no signs that the person understands what others are expressing.

C6. Expressive communication extension. Estimate the number of words, gestures, signs, objects of reference etc. that are actively used by the person (mark one alternative)

- 1) Normal for age
- 2) Above 100 signs, words, objects of reference etc. but not normal for age
- 3) Between 21 to 100 gestures, signs, words, objects of reference etc.
- 4) Between 1 to 20 gestures, signs, words, objects of reference etc.
- 5) No or almost no expressive communication

After having filled in the information above, discuss and describe the consequences for education and non-medical service provision, and recommendations for interventions.

Consequences of language loss

For persons with severe visual impairment, social interaction is mainly based on language use. Although individuals with JNCL develop severe language problems, language continue to be an important tool for social participation and well-being. Comments in the present study indicate that the need for and wish to be in interaction and dialogue with other people are not reduced by the disease. Young people with JNCL may become frustrated by not having the ability to participate socially in the same way as they used to. Their communicative initiations are often frequent despite pervasive speech problems. The loss of speech and reduction in comprehension cause significant frustration and stress in both the young person with JNCL and in the family (von Tetzchner, Fosse, & Elmerskog, 2013). Persons who have problems with communication and language – irrespective of diagnosis – often experience misunderstandings and frustration (von Tetzchner & Martinsen, 2000). People with JNCL experience the additional strain of losing skills that they earlier mastered with ease.

During the course of the disease, people with JNCL become increasingly dependent on support. Many will need assistance from others to communicate successfully and participate in social activities. Conversations become more tiresome and frustrating, both for the person with JNCL and the communication partners. These difficulties will affect the person's social life and total life situation and make spontaneous communication with peers and others difficult. The problems with language and social interaction may create frustration and elicit despair, anger and other emotional reactions (see Chapter 27). They want to communicate as much as before but do not have the ability to participate in the same way as they used to.

When the parents in the present study were asked to evaluate the extent of the negative social consequences of speech problems on a scale from 1 (no negative effect) to 5 (high negative effect) (see Appendix A), 43 percent of the parents answered that speech problems had a high or very high negative effect on social life. No parents answered that the problems with speech did not have any negative effect on social life.

The central role of language and communication in all aspects of life indicates that research should give priority to developing interventions that may compensate for the deterioration of language by strengthening or at least supporting the existing communication skills for as long as possible.

Any discussion of intervention must address the need to choose and implement measures in advance of symptom development to make the best possible foundation for meeting future declines. Of necessity, when symptoms are evident – one must also decide what to do next. The goal is to prolong the period with fluent communication for the person with JNCL. The EDO can potentially be a helpful tool when making decisions for today and for the future.

References

- Bang, J., Spina, S., & Miller, B. L. (2015). Frontotemporal dementia. *The Lancet*, 386 (10004), 1672–1682.
- Bates, E., Bretherton, I., & Snyder, L. S. (1988). *From first words to grammar: Individual differences and dissociable mechanisms*. Cambridge, UK: Cambridge University Press.
- Bauman-Waengler, J. (2016). *Articulatory and phonology in speech sound disorders: A clinical focus, Fifth edition*. Boston, MA: Allyn and Bacon.
- Bendixen, A. (1996). Hele livssituasjonen vår er snudd opp ned (Our life situation is turned upside down). Master thesis, University of Trondheim.
- Best, W. (2005). Investigation of a new intervention for children with word-finding problems. *International Journal of Language and Communication Disorders*, 40, 279–318.
- Bishop, D. V. M. (2006). What causes specific language impairment in children? *Current Directions in Psychological Science*, 15, 217–221.

- Bloom, L. (1974). Talking, understanding, and thinking: Developmental relationship between receptive and expressive language. In R.L. Schiefelbusch & L. L. Lloyd (Eds.), *Language perspectives, acquisition, retardation and intervention* (pp. 285–311). Baltimore, MD: University Park Press.
- Chaffey, A. M. (1987). Avvik i setninger med negasjon hos en pasient med Spielmeyer-Vogts sykdom (Deviations in sentences with negation in a patient with Spielmeyer-Vogt disease). *Arbeidsskrift for Nordisk Språk og Litteratur*, 52, 129–140.
- Elmerskog, B., & Fosse, P. (Eds.) (2012). *Lære for livet. En innføring i pedagogikk og læring ved juvenil neuronal ceroid lipofuscinose (JNCL)* (Learning for life. An introduction to education and learning in juvenile neuronal ceroid lipofuscinosis (JNCL). Trondheim: Akademika forlag.
- Erin, J. N., & Koenig, A.J. (1997) The student with a visual disability and a learning disability. *Journal of Learning Disabilities*, 30, 309–320.
- Fraiberg, S. (1977). *Insights from the blind*. New York: Basic Books.
- Gayton, R. (1982). The education and care problems of visually handicapped children suffering from juvenile JNCL. Master's thesis, University of Birmingham.
- Goldman, D., & Fristoe, M. (2015). *Goldman-Fristoe Test of Articulation, Third edition (GFTA-3)*. Circle Pines, MN: American Guidance Service.
- Gombault, E. (Ed.) (2010). *Pädagogische Förderung und Begleitung von Kindern und Jugendlichen mit juveniler Neuronaler Ceroid-Lipofuscinose* (Educational requirements and support of children and young persons with JNCL). Hamburg: Bildungszentrum für Blinde und Sehbehinderte.
- Graven, T. (2018). How individuals who are blind locate targets. *British Journal of Visual Impairment*, 36, 57–74
- Grove, N., Bunning, K., Porter, J., & Olsson, C. (1999). See what I mean: Interpreting the meaning of communication by people with severe and profound intellectual disabilities. *Journal of Applied Research in Intellectual Disabilities*, 12, 190–203.
- Hyltenstam, K., & Viberg, Å. (1993). *Progression and regression in language: sociocultural, neuropsychological and linguistic perspectives*. Cambridge: Cambridge University Press.
- Klimova, B., & Kuca, K. (2016). Speech and language impairments in dementia. *Journal of Applied Biomedicine*, 14, 97–103.
- Kohlschütter, A., Laabs, R. & Albani, M. (1988). Juvenile neuronal ceroid lipofuscinosis (SV disease) : Quantitative description of its clinical variability. In P. Uvebrant & H. Svensson (Eds.), *Proceedings of the Nordic Meeting on Spielmeyer-Vogt's disease* (pp. 4–12). Almåsa: The Swedish Federation of the Visually Handicapped.
- Laabs, R. (1988). Zur klinischen Variabilität der Juvenilen Neuronalen Ceroid-Lipofuscinose. Versuch einer quantitativen Beschreibung des Verlaufs bei 17 Patienten (On the clinical variation in juvenile neuronal ceroid lipofuscinosis. An attempt to quantitative description of the course of 17 patients). Doctoral thesis, University Hospital, Eppendorf, Germany.
- Landau, B., & Gleitman, L.R. (1985). *Language and experience: Evidence from the blind child*. Cambridge, MA: Harvard University Press.
- Lund, N. J., & Duchan, J. F. (1993). *Assessing children's language in naturalistic contexts, Third edition*. Englewood Cliffs, NJ: Prentice-Hall.
- Messer, D., & Dockrell, J. E. (2006). Children's naming and word finding difficulties: descriptions and explanations. *Journal of Speech, Language and Hearing Research*, 49, 309–332.
- Pérez-Pereira, M., & Conti-Ramsden, G. (1999). *Language development and social interaction in blind children*. Hove, UK: Psychology Press.

- Pijnacker, J., Vervloed, M. P., & Steenbergen, B. (2012). Pragmatic abilities in children with congenital visual impairment: An exploration of non-literal language and advanced theory of mind understanding. *Journal of Autism and Developmental Disorders*, *42*, 2440–2449.
- Rohrer, J. D., Knight, W. D., Warren, J. E., Fox, N. C., Rossor, M. N., & Warren, J. D. (2008). Word-finding difficulty: A clinical analysis of the progressive aphasia. *Brain*, *131*, 8–38.
- Tadic, V., Pring, L., & Dale, N. (2010). Are language and social communication intact in children with congenital visual impairment at school age? *Journal of Child Psychology and Psychiatry*, *51*, 696–705.
- Tomasello, M. (2006). Acquiring linguistic constructions. In W. Damon, R. M. Lerner, D. Kuhn & R. S. Siegler (Eds.), *Handbook of child psychology, Sixth edition, Volume 2: Cognition, perception and language* (pp. 371–384). New York, NY: Wiley.
- van Delden, G. W. (2009). Education for children with Batten disease. Presented at 7th European Conference of ICEVI, Dublin, Ireland, 2009.
- Velleman, S. L. (2016). *Speech sound disorders*. Philadelphia, PA: Lippincott, Williams and Wilkins.
- von Tetzchner, S. (1993). *Utviklingen hos mennesker med Spielmeier-Vogt-syndromet med spesiell vekt på språklige og kognitive funksjoner* (Development in individuals with Spielmeier-Vogt syndrome with emphasis on language and cognitive functioning). Melhus, Norway: Tambartun Resource Centre for the Visually Impaired.
- von Tetzchner, S. (2006). Communication and language in Batten disease. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 101–103) Örebro, Sweden: Swedish Institute for Special Needs Education.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- von Tetzchner, S., Fosse, P., Elmerskog, E. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta*, *1832*, 1894–1905.
- von Tetzchner, S. & Martinsen, H. (2000). *Introduction to augmentative and alternative communication*. London: Whurr/Wiley.

Motor Development and Loss in Individuals with Juvenile Neuronal Ceroid Lipofuscinosis

Bengt Elmerskog and Riikka Hokkanen

Physical actions are voluntary movements with intentions and goals, which allow humans to overcome gravitational forces, to plan, coordinate, perform and evaluate actions, and to create new physical and social opportunities for action. The ability to locomote is a core element of human adaptation and motor skills develop through maturation and practice (Hadders-Algra, 2018; Piaget, 1983). Individual differences may originate in biological differences as well as environmental and cultural factors (Adolph & Robinson, 2015; Houwen, Visscher, Lemmink, & Hartman, 2009). Children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL) show gradual changes in motor abilities and activity. These changes appear to be related to motor decline caused by the disease but also to visual impairment and emergent dementia.

Motor functions and impairment

Gross motor skills involve the large muscle groups, such as body posture and walking, balancing, climbing, swimming and crawling. Independent locomotion is important for action and participation. Gross motor skills enable exploration of objects, people and places, and thus influence the whole psychology of the child (Anderson, 2018; Anderson et al., 2014; Campos et al., 2000). The basic gross motor skills develop during early childhood but the quality of movements and motor coordination continue to improve into young adulthood. When gross motor skills are established, they are usually maintained even after longer periods of non-use. For example, walking and biking do not require much practice once they have been learned and mastered (Galahue & Ozmun, 2006; Stallings, 1973).

Fine motor skills involve the use of smaller muscle groups and make it possible to manually handle and explore things. They include movements of the wrists, hands and fingers, for example for pointing, gripping, reading braille or Moon,

playing the piano, dressing and painting. Many fine motor skills are present in early childhood but fine motor skills continue to develop into young adulthood. Fine motor skills require more exercise than gross motor skills to be maintained, they will to a larger extent be lost if they are not exercised. For example, playing the piano on a high level requires continuous exercises (Stallings, 1973).

Physical activity usually has associated health benefits. Gross motor activities are influenced by the individual's physical condition and health, the strength and endurance of muscles and the uptake of oxygen. They support health because they require substantial energy, while fine motor activities do not confer the same health benefits. One's physical condition and motor abilities may mirror one's lifestyle, reflecting whether muscles are exercised on a regular basis (Stallings, 1973; Tremblay, Colley, Saunders, Healy, & Olsen, 2010; Van Duyn et al., 2007).

Physical impairment may be caused by a bodily abnormality or a neurological condition that causes partial or total loss of the function of one or more body parts, such as movements of the limbs. Physical impairment can for example cause muscle weakness, poor endurance, lack of muscle control, reduced co-ordination, or total paralysis. Physical impairments are evident in neurological conditions such as cerebral palsy, muscular dystrophy, Parkinson disease and JNCL. An extreme form of physical impairment is the locked-in syndrome, where the individual's control of muscles is lost while cognitive functions are not affected by the disease (Haig, Katz, & Saghal, 1987). Developmental coordination disorder (dyspraxia) is a neurological condition that affects the person's ability to plan and coordinate physical movements, balance, posture and motor actions, but does not involve paralysis or motor impairment in the conventional sense (Gibbs, Appleton, & Appleton, 2007).

A loss or decline of motor function can consequently be influenced by bodily and neurological motor impairment, while reduced practice will impede learning, development or maintenance of motor skills. A sedentary lifestyle will contribute to reduced motor skills. To promote the best possible motor function, education should focus on skills in the zone of proximal development (i.e., those that a child is on the way to master) or, if the child is showing regression, on skills within the zone of developmental maintenance (i.e., skills that can be maintained with adaptation and help) (see Chapter 15). This focus will also support the best possible health in children, adolescents and adults with JNCL.

Being physically active does not come easily when children and young people have severe motor impairments. People with physical impairments are often not sufficiently supported – or even hindered – in participating in physical activities, due to lack of environmental adaptations and individual support (Gibbs, Brown, & Muir, 2008). Severe motor disabilities make performance of aerobic exercises difficult, that is, exercises that make one perspire and breathe harder, and the

heart to beat faster (Læssøe, 2011). Even moderate impairments tend to imply a sedentary lifestyle and limited social and societal participation, which again affect the person's physical condition and health. Severe motor impairments may thus have a negative effect on development in general, health and participation.

Physical barriers in the environment may represent significant hindrances to being physically active for people using wheelchairs, but also for people with coordination or balance problems. Steps, curbs or a rough ground make independent movement not just difficult, but aversive if falls occur, and should be removed or adapted when possible and required. Physical play and sport activities, such as climbing or playing soccer, can contribute positively to motor development. Such activities are usually not accessible for children with severe motor disorders (Gibbs et al., 2008).

Minimal use of some gross motor movements may in addition have a negative effect on the joints and the tendons attached to the muscles. A joint that is not exercised or maintained over time will become stiffer and the range of movement in the joint will become reduced or even arrested. Contractures (permanent shortening of muscles, tendons, or ligaments) may entail pain when performing daily activities such as dressing or change of position in bed (Jeremiassen, 2016).

Decline of motor function and activities in individuals with JNCL

Most children with JNCL have typical motor development in the early years. At the time of diagnosis, children with JNCL can walk, jump, and run normally, and they do not have any problems with fine motor skills. However, with age, abnormalities in gait and slight reductions in voluntary coordination of muscle movements and motor functions controlling speech become noticeable (Østergaard, 2016) (see Chapter 6). Some children with JNCL develop an extrapyramidal motor disorder that resembles Parkinson disease around the age of 10 to 12 years (Järvelä, 1997), with hypokinesia (reduced bodily movement), rigidity of muscles and joints, shuffling gait, reduced balance and a stooped posture (see Figure 3.3 in Chapter 3). In a Finnish study of 53 persons with JNCL, the average age at the first sign of such Parkinsonian type of walking was 13.7 years (Santavuori, Heiskala, Westermarck, Saino, & Moren, 1988). Some of the symptoms can sometimes be temporarily ameliorated with anti-Parkinson medication (Åberg, Rinne, Rajantie, & Santavuori, 2001).

JNCL affects both gross and fine motor activities. In the present study (Appendix A), parents were asked when they first observed problems with gait and use of the hands and arms. Figure 7.1 shows that for most participants,

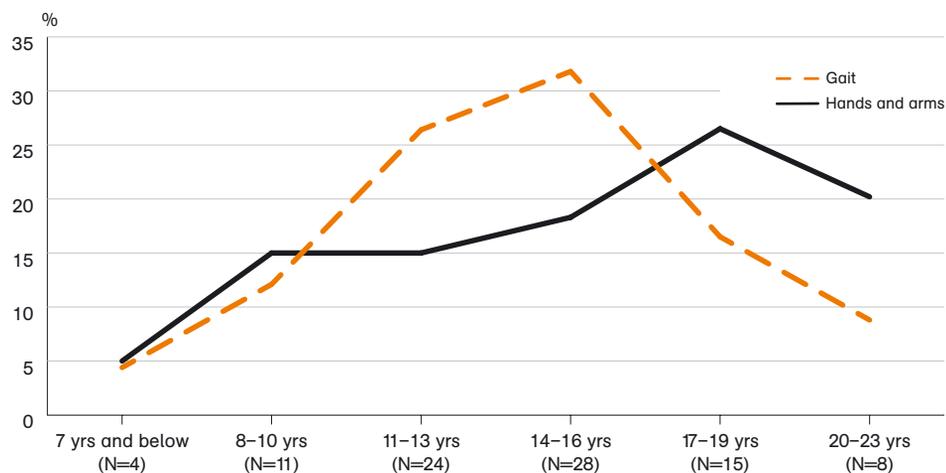


Figure 7.1 Percentage of children who first showed problems with gait or hands and arms at the different age intervals

gross motor problems appeared earlier than fine motor problems. Gross motor problems often appeared in early adolescence, gait problems were first observed at an average age of 14.1 years, while problems related to the use of the hands and arms typically appeared in the late teens (average 15.5 years).

The later stages of the disease may include increased rigidity, slowness of movement such as slow steps with flexion in hips and knees, a shuffling gait with shortened steps, reduced arm swings and a forward-flexed posture. Involuntary movements are also reported. Many persons with JNCL need a wheelchair in late adolescence or early adulthood but there is considerable variation in the group (Østergaard, 2016, 2018). Not all persons with JNCL lose the ability to walk. A Finnish study found an average age of 17.3 years for losing the ability to walk (Järvelä, Autti, Lamminranta, Åberg, Raininko, & Santavuori, 1997). A Danish study found that females became dependent on a wheelchair at the average age of 17 years (range 13.8–20.6), while males became dependent on a wheelchair at 20.2 years (range 14.3–22.1) (Nielsen & Østergaard, 2013). Motor dysfunctions include chewing and swallowing difficulties and may be observed from the late teens. To ensure adequate intake of nourishment, feeding with a gastric tube may be required (Østergaard, 2016).

The results indicate that problems with upholding a healthy lifestyle due to motor impairment are likely to emerge around 11 to 13 years in young people with JNCL, unless met by appropriate measures. In the present study, a few individuals with JNCL were able to walk long distances around the age of 25 years. They showed no evidence of extrapyramidal motor disorder, abnormalities in rigidity,

or chewing and swallowing difficulties, and also had no major difficulties in the performance of fine motor skills. The variation in the onset of decline in fine and gross motor skills thus seems to be considerable in individuals with JNCL.

Visual impairment

Visual impairment will usually influence participation in motor activities, and severe visual impairment may entail a sedentary life style. Sight is important for exploring the world from a distance and visual observations motivate exploration of places and objects and participation in physical activities like moving around, participating in sports, and for stimulating us to learn and refine motor skills, for instance by observing others (Fraiberg, 1977). For instance, children who are blind may not be aware of a red ball on the floor and thus not start playing with it or experience the thrill that comes from noticing a tree in the garden that invites climbing. Lacking incentives to explore the surroundings are likely to have negative influences on motor activities and development in children who are blind.

Children who are blind move less to explore, and their motor skills develop later due to a lack of visual incentives for movement (Freedman & Cannady, 1971). Moreover, in children with congenital or very early-acquired blindness there are significant structural changes in the brain, which may be beneficial for attention and acting, but they also imply some maladaptation to the sensory information that is accessible (Singh, Phillips, Merabet, & Sinha, 2018). This does not happen to the same extent when visual impairment starts later. There are thus significant differences in neurological and adaptive resources between the developmental courses of individuals with congenital or early onset of blindness, and individuals who lose their sight in middle childhood or later.

Individuals with JNCL, who start to lose sight during preschool years and become blind in middle childhood, have mainly used visual cues for orienting and continue to do so after the visual problems appear. Yet, they may underperform and function more like sighted people who are blindfolded (Schinazi, Thrash, & Chebat, 2016; see Chapter 16). The link between loss of vision and loss of motor skills needs to be considered. Children and young people with JNCL can no longer rely on the perceptual cues that they used earlier in life. Even if met with appropriate measures, they may also start to lose some motor skills due to lack of incentive as well as increased insecurity.

Students who are blind need more cognitive resources and learning to orientate themselves than do sighted peers (see Chapter 16). A Norwegian study found that cognitive-based orientation was difficult for blind children before the motor actions were automatized, and concludes that problems in orientation may have a negative

influence on motor performance in this group (Tellevik, Storliløkken, Martinsen, & Elmerskog, 2007). Performing daily activities demands more energy for people who are blind and the need to be fit might even be greater (Buell, 1984). Poor posture due to a sedentary lifestyle is relatively common among blind children and persons with severe visual impairment with no other disabilities they tend to show a lower level of fitness than sighted peers. They have a higher rate of obesity, higher degree of muscle weakness and lower cardiovascular endurance than peers of same age (Aslan, Calik, & Kitis, 2012; Lieberman & McHugh, 2001; Weil, Wachterman, McCarthy, Davis, O'Day, Iezzoni, & Wee, 2002). In addition, children with severe visual impairments are often not expected to pursue a full range of life goals (Lieberman & McHugh, 2001). The lack of expectations among people in the environment may also lead to reduced motor activity and furthermore to obesity and other health risk factors (Weil et al., 2002).

Children who are blind are more dependent on adult helpers than their peers who are sighted. This reliance in turn may create another barrier for children's engagement in motor activities; adults' preferences and priorities do not always align with those of children. The adult world is formed by preplanned schedules, movements are done with a high degree of time-efficiency, and adults will in many cases avoid situations where they become dirty and wet, which frequently characterize the explorative activities that usually are stimulating for children and young people. Children who are blind and their adult guides will often move in accordance with the adult culture, taking the shortest and most efficient way between two locations. However, this need for time efficiency also reduces the total amount of physical activity for everyone (Elmerskog & Fosse, 2013).

The difference in movement and physical activity between children who are blind and children who are sighted may be considerable, affecting the long-term physical status, gross motor development, and the personal development of children who are blind (Houwen et al., 2009; Elmerskog et al., 1993; Fraiberg, 1977). There is a significant risk that early blindness may lead to a sedentary lifestyle. Such a lifestyle should be prevented to support physical and mental health, through appropriate adaptation of physical activities, guidance and support (Chapala et al., 2017).

Effect of motor and visual impairments on activities

Children and adolescents with severe visual impairment are often encouraged to focus on activities requiring fine motor skills (Cheadle, 1994) which make small demands on orientation ability (see chapter 16). Many individuals with congenital blindness become good at playing the piano, reading braille or dressing, which mainly require the use of hands and arms.

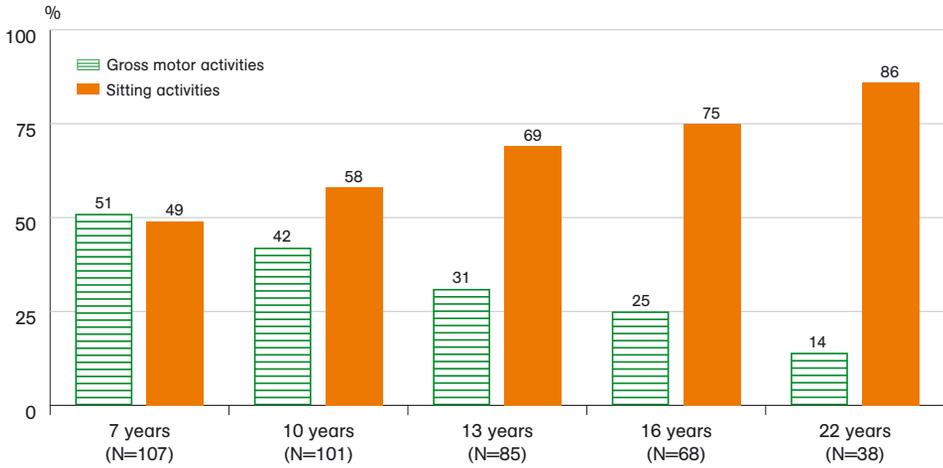


Figure 7.2 Percentage of gross motor and sitting activities as main activities at different age intervals

In the present study (Appendix A), the parents were asked what kind of activities their children were mostly occupied with at different age spans. Figure 7.2 shows that the relative participation in gross motor activities decreased gradually from age seven to twenty-two years compared to sitting activities. There is no doubt that the reduced physical activity and increased passivity in early life are related to the visual impairment but also other factors, such as needs for increased

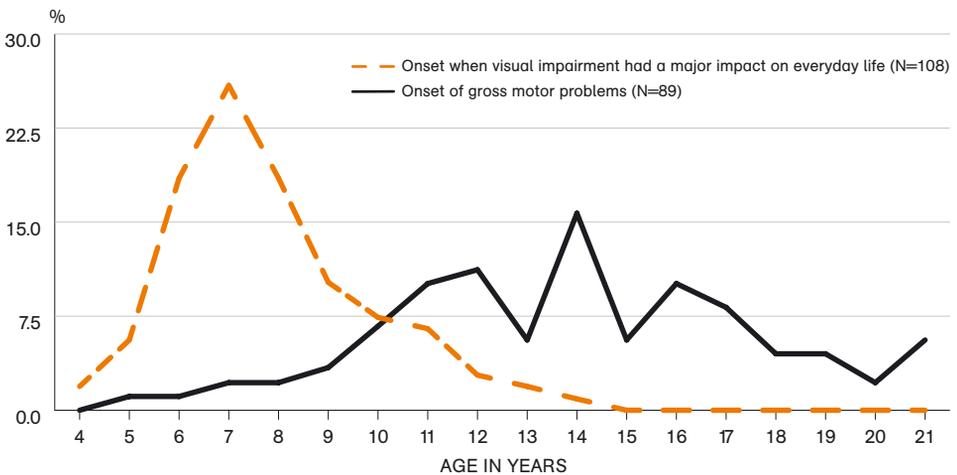


Figure 7.3 Percentage of participants with onset of visual impairment of major impact and onset of gross motor problems at different age levels

support and adaptations in the environment. Figure 7.3 shows the age when the visual impairment had a major impact on the participants' functioning and the age of onset of gross motor problems. The visual decline had begun to have a major impact on everyday life at an average age of 7.2 years among the participants, while total blindness occurred around the age of 10.7 years (see Chapter 4). Onset of gross motor decline appeared significantly later, around 14.2 years on average. There are reasons to believe that the early decreases in participation in gross motor activities among individuals with JNCL have other causes than the later declines in motor skills.

The participants in the present study were on average affected by a severe visual impairment between age 6 and 9 years (see Chapter 4). The initial physical inactivity thus seems to be related to visual decline rather than loss of motor functions. Visual impairment affects agility, speed, balance, orientation, independence and so forth. The participants stopped taking part in gross motor activities like playing soccer, climbing trees and playing involving gross motor actions as a consequence of the visual impairment. Most of the gross motor activities that children between the age of 7 and 13 years participate in are heavily dependent on vision, and these are difficult to perform for children with JNCL. The reduction in participation in gross motor activities shown in Figure 7.2 reflects a change in lifestyle forced by the visual impairment. The following quotation from a parent illustrates this:

Our daughter of 11 years of age stopped doing physical activities when she became blind three years ago. She was more or less exempted from doing P.E. with others at school, there were no alternatives for her to participate in sport activities in our town. She prefers doing smooth activities, like singing, listening to music or audio books or playing with her dolls (she also loves activities like drawing). She became physically inactive, it is difficult for us to encourage her to do physical activities, partly because she cannot share such activities with her sighted friends.

Children with JNCL require adaptations of accessible gross motor activities and the environment, and support from adults. Schools have an important role in promoting and supporting a physically active lifestyle in these students. In the present study, many of the participants first attended mainstream schools that had limited experience with students who have visual impairment and little insight into how this disability may affect the students' daily living (see Chapter 9). Many schools make good efforts to promote the students' academic performance but the comments in the surveys and interviews did not indicate that the schools were initiating measures to prevent or meet the students' growing sedentary lifestyle,

for example with proactive, precautionary or enhanced teaching (see Chapters 10 and 12). For instance, almost none of the parents and staff reported that extra resources were used at school for adapted physical education in early school age.

There are thus reasons to believe that the early decline in participation in gross motor activities often reflects a life-style problem related to the visual problems. This emphasizes the importance of distinguishing between an impairment and the developmental consequences of the impairment, consequences which are influenced by a range of other factors (von Tetzchner, 2019). The motor impairment (and possibly other disorders) that emerges as JNCL progresses increases the decline in participation in activities involving gross motor skills. The results in Figure 7.3 indicate that promoting a best possible motor development and maintenance plan for children and young people with JNCL requires a multidisciplinary approach, in particular between specialists with competence in visual impairment and motor impairment.

Physical activity, cognition and childhood dementia

There is a general positive association between an active physical lifestyle and cognitive functioning. Physical activity contributes positively to maintain functions in elderly people with mild and moderate dementia (see Chapter 5). There is therefore interest in the potential impact that interventions leading to improved fitness and exercise might have on the maintenance of cognitive function and learning in persons with JNCL.

Physical activity and cognition

A number of studies have observed a positive influence of physical exercise on cognitive functioning and academic performance in both children and adults without dementia (Donnelly et al., 2016; Sofi et al., 2011). For example, a sedentary lifestyle was linked to poorer reading skills in the first three school years in 6 to 8 years old boys according to a study from Finland (Haapala et al., 2017). Results from other studies indicate that higher levels of activity or fitness enhance thinking, concentration and academic performance, and students with good fitness obtained higher scores on standardized tests of cognition and brought home better report cards (Coe, Pivarnik, Womack, Reeves, & Malina, 2006; Coe, Peterson, Blair, Schutten, & Peddie, 2013). Coe and associates conclude that there is enough evidence to support a greater provision of physical activity into the school curriculum. Other studies have found that young adults who run or

participate in other aerobic activities maintain memory and other cognitive skills better in middle age. For instance, one study found that young adults (on average 25 years old) with a physically active life style, who were running or participating in aerobic activities, showed better cognitive functioning about 25 years later (age 43 to 55 years) (Zhu et al., 2014).

The assumption is that physical activity influences the brain (Zhu et al., 2014). One explanation is that moderately intense exercise can increase the size of the hippocampus, an area of the brain involved in learning and memory, which in turn leads to better thinking and problem-solving, more focused attention and improved learning. It is the endorphins the brain releases during exercises that help children to improve mood, energy levels and even sleep. It is argued that physical activity and exercise are significant for children's health, wellbeing and development, and that physical activities should always be considered when promoting children's development, in particular in children with illnesses and disabilities.

An active physical life thus seems to promote learning and cognition in children and adults, although many factors influence cognition and learning (Wang, Xu, & Pei, 2012), and the statistical effects of physical activity are small or moderate (Donnelly et al., 2016; Keeley & Fox, 2009; Singh, Uijtewilligen, Twisk, van Mechelen, & Chinapaw, 2012; Tomporowski, Davis, Miller, & Naglieri, 2008).

Physical activity and dementia

Physical activity seems to both reduce the risk for dementia and contribute to maintaining cognitive functioning in elderly people with mild and moderate dementia (Aarsland et al., 2010; Sofi et al., 2011). A Finnish twin study found that physical activity in midlife (age 24–60) seemed to decrease the risk of dementia in old age, 29 years later. The participants who were engaged in leisure-time physical activity at least twice per week had a lower risk of dementia than individuals who were less active (Iso-Markku et al., 2016). Other studies present similar findings, that adults with an active physical lifestyle have lower risk for Alzheimer disease and other forms of dementia than less physically active persons (Laurin, Verreault, Lindsay, MacPherson, & Rockwood, 2001; Rovio et al., 2005).

Most studies have compared people with different life styles across the lifespan but there is also some evidence that intervention with physical activity in elderly people may be beneficial. In one study, elderly people who had memory problems and were at risk for Alzheimer disease were allocated to an intervention group who did 20 minutes of extra physical exercise every day or a control group who did not do any extra physical activity. After six months, there was a small improvement in performance on cognitive tasks in the intervention group compared to the control group, and the difference was maintained 12 months after

the experimental intervention was stopped (Lautenschlager et al., 2008). There is thus evidence that increasing physical activity also later in life reduces the risk for dementia (Lautenschlager, Cox, & Cyarto, 2012).

Dementia, physical activity and JNCL

The studies cited above indicate that physical activities to some extent can prevent cognitive decline and dementia among elderly and middle-aged people. Childhood dementia is a core characteristic of JNCL (see Chapter 5) but a literature search for research about childhood dementia and physical activities yielded no results, probably due to the low prevalence of such diseases. Professionals and lay people have rarely heard about JNCL and childhood dementia, and this seems to be the case even for professionals in special education and social services. Moreover, the relevance of research evidence from elderly adults has not yet been acknowledged by the educational and social services that are responsible for children, adolescents and young adults with JNCL. However, it is likely that physical activity and an active lifestyle will have similar positive effects on cognition, learning and well-being in persons with childhood dementia, including persons with JNCL. The following quotation supports such an assumption:

My son with JNCL is 16 years old. He lived a rather inactive life until he was 14. We noticed this was not good for him. His physical condition was low, it was difficult to engage him in activities, he showed low spirits close to depression, and his behavior changed from day to day. It was not easy to deal with him. We decided to involve him in physical activities on a daily basis. After some struggle in the beginning he learned to love it. Today we do physical exercises one hour per day. He asks for the activities, his mood has improved and is more stable than before, and he is actually doing better at school.

Even if this is the experience of just one young person and his family, it seems important to follow it up in research and practice.

Conclusions

Both visual impairment and motor impairment will make participation in a healthy lifestyle more difficult. Individuals with JNCL typically become visually impaired in the early school years, while gross motor problems become observable in early

teens and fine motor problems in the later teens. However, there are considerable differences in how and when impairments appear in individuals with JNCL. The disease causes loss of motor skills in individuals with JNCL, but motor performance is also influenced by lifestyle and factors in the environment. Education cannot heal or stop the progress of JNCL but can certainly affect the individual's lifestyle and participation. On the basis of research with children and adults with typical development, as well as adults with mild and moderate dementia, and comments in the present study, it seems justified to suggest that a physically active lifestyle should be a mandatory goal and integrated into Individual Education Plans and Habilitation Plans for all individuals with JNCL, from childhood to adulthood. Promoting a healthy lifestyle entails a focus on compensation and should proceed in accordance with the principles of proactive, precautionary and enhanced learning and stimulation (see Chapters 11 and 12).

References

- Aarsland, D., Sardahaee, F. S., Anderssen, S., Ballard, C., & the Alzheimer's Society Systematic Review group. (2010). Is physical activity a potential preventive factor for vascular dementia? A systematic review. *Aging and Mental Health, 14*, 386–395.
- Åberg, L. E., Rinne, J. O., Rajantie, I., & Santavuori, P. (2001). A favorable response to antiparkinsonian treatment in juvenile neuronal ceroid lipofuscinosis. *Neurology, 56*, 1236–1239.
- Adolph, K. E., & Robinson, S. R. (2015). Motor development. In R. M. Lerner, L. Liben & U. Muller (Eds.), *Handbook of child psychology and developmental science, Seventh edition, Volume 2: Cognitive processes* (pp. 114–157). New York, NY: Wiley.
- Anderson, D. I. (2018). Motor development: Far more than just the development of motor skills. *Kinesiology Review, 7*, 99–114.
- Anderson, D. I., Campos, J. J., Witherington, D. C., Dahl, A., Rivera, M., He, M., et al. (2013). The role of locomotion in psychological development. *Frontiers in Psychology, 4*, 440.
- Aslan, U. B., Calik, B. B., & Kitiş, A. (2012). The effect of gender and level of vision on the physical activity level of children and adolescents with visual impairment. *Research in Developmental Disabilities, 33*, 1799–1804.
- Buell C. E. (1984). Physical education for blind children. *British Journal of Visual Impairment, 2*, 107–122.
- Campos, J. J., Anderson, D. I., Barbu-Roth, M. A., Hubbard, E. M., Hertenstein, M. J., & Witherington, D. (2000). Travel broadens the mind. *Infancy, 1*, 149–219.
- Cheadle, B. (1994). A parent's guide to the slate and stylus. *Future Reflections, 13*, 6–14
- Coe, D. P., Peterson, T., Blair, C., Schutten, M. C., & Peddie, H. (2013). Physical fitness, academic achievement, and socioeconomic status in school-aged youth. *Journal of School Health, 83*, 500–507.
- Coe, D. P., Pivarnik, J. M., Womack, C. J., Reeves, M. J., & Malina, R. M. (2006). Effect of physical education and activity levels on academic achievement in children. *Medicine and Science in Sports and Exercise, 38*, 1515–1519.

- Donnelly, J. E., Hillman, C. H., Castelli, D., Etnier, J. L., Lee, S., Tomporowski, P., Lambourne, K., & Szabo-Reed, A. N. (2016). Physical activity, fitness, cognitive function, and academic achievement in children: A systematic review. *Medicine and Science in Sports and Exercise*, *48*, 1197–1222.
- Elmerskog, B., & Fosse, P. (Eds.) (2012). *Lære for livet: en innføring i pedagogikk og læring ved juvenil neuronal ceroid lipofuscinose (JNCL)* (Learning for life. An introduction to education and learning in juvenile neuronal ceroid lipofuscinosis). Trondheim: Akademika forlag.
- Elmerskog, B., Martinsen, H., Storliløkken, M., & Tellevik, J. M. (1993). *Førhøretssopplæring – Mobility i en funksjonell sammenheng* (Mobility training in a functional context). Trondheim: Tapir.
- Fraiberg S., 1977. *Insights from the blind*. New York: Basic Books.
- Freedman, D. A., & Cannady, C. (1971). Delayed emergence of prone locomotion. *Journal of Nervous and Mental Disease*, *153*, 108–117.
- Gallahue, D. L., & Ozmun, J. C. (2006). *Understanding motor development: Infants, children, adolescents, adults, Sixth edition*. Boston, MA: McGraw-Hill.
- Gibbs, J., Appleton, J., & Appleton, R. (2007). Dyspraxia or developmental coordination disorder? Unravelling the enigma. *Archives of Disease in Childhood*, *92*, 534–539
- Gibbs, S. M., Brown M. J., & Muir W. J. (2008). The experiences of adults with intellectual disabilities and their careers in general hospitals: A focus group study. *Journal of Intellectual Disability Research*, *52*, 1061–1077.
- Haapala, E. A., Väistö, J., Lintu, N., Westgate, K., Ekelund, U., Poikkeus, A. M., Brage, S., & Lakka, T. A. (2017). Physical activity and sedentary time in relation to academic achievement in children. *Journal of Science and Medicine in Sport*, *20*, 583–589.
- Hadders-Algra, M. (2018). Early human motor development: From variation to the ability to vary and adapt. *Neuroscience and Biobehavioral Reviews*, *90*, 411–427.
- Haig, A. J., Katz, R. T., & Sahgal, V. (1987). Mortality and complications of the locked-in syndrome. *Archives of Physical Medicine and Rehabilitation*, *68*, 24–27.
- Houwen, S., Visscher, C., Lemmink, K. A. M., & Hartman, E. (2009). Motor skill performance of children and adolescents with visual impairments: A review. *Exceptional Children*, *75*, 464–492
- Iso-Markku, P., Waller, K., Vuoksima, E., Heikkilä, K., Rinne, J., Kaprio, J., & Kujala, U. M. (2016). Midlife physical activity and cognition later in life: A prospective twin study. *Journal of Alzheimer's Disease*, *54*, 1303–1317.
- Järvelä, I., Autti, T., Lamminranta, S., Åberg, L., Raininko, R., & Santavuori, P. (1997). Clinical and magnetic resonance imaging findings in batten disease: Analysis of the major mutation (1.02-kb deletion). *Annals of Neurology*, *42*, 799–802.
- Jeremiassen, R. E. H. (2016). *JNCL og fysisk aktivitet* (JNCL and physical activity). Trondheim: Statped.
- Keeley, T. J., & Fox, K. R. (2009). The impact of physical activity and fitness on academic achievement and cognitive performance in children. *International Review of Sport and Exercise Psychology*, *2*, 198–214.
- Laurin, D., Verreault, R., Lindsay, J., MacPherson, K., & Rockwood, K. (2001). Physical activity and risk of cognitive impairment and dementia in elderly persons. *Archives of Neurology*, *58*, 498–504.
- Lautenschlager, N. T., Cox, K. L., Flicker, L., Foster, J. K., van Bockxmeer, F. M., Xiao, J., Greenop K. R., & Almeida, O. P. (2008). Effect of physical activity on cognitive function in older adults at risk for Alzheimer disease: A randomized trial. *Jama*, *300*, 1027–1037.
- Lautenschlager, N. T., Cox, K., & Cyarto, E. V. (2012). The influence of exercise on brain aging and dementia. *Biochimica et Biophysica Acta (BBA) – Molecular Basis of Disease*, *1822*, 474–481.

- Læssøe, U. (2011). Forudsætninger for bevægelse: kroppsfunktionsniveau (Prerequisites for movement: body function level. In A.-M. Kissow, H. Lisby & U. Læssøe (Eds.), *Bevegelse: En grundbog* (pp. 83–102). Copenhagen, Denmark: Munksgaard.
- Lieberman, L., & McHugh, E. (2001). Health-related fitness of children who are visually impaired. *Journal of Visual Impairment and Blindness*, *95*, 272–287.
- Nielsen, A. K., & Østergaard, J. R. (2013). Do females with juvenile ceroid lipofuscinosis (Batten disease) have a more severe disease course? The Danish experience. *European Journal of Paediatric Neurology*, *17*, 265–268.
- Ostergaard, J. R. (2016). Juvenile neuronal ceroid lipofuscinosis (Batten disease): Current insights. *Autonomic Neuroscience*, *214*, 73–83.
- Ostergaard, J. R. (2018). Paroxysmal sympathetic hyperactivity in Juvenile neuronal ceroid lipofuscinosis (Batten disease). *Autonomic Neuroscience*, *214*, 15–18.
- Piaget J. (1983). Piaget's theory. In P. Mussen (Ed.), *Handbook of child psychology, Fourth edition, Volume 1: History, theory and methods* (pp. 103–128). New York: Wiley.
- Rovio, S., Kåreholt, I., Helkala, E. L., Viitanen, M., Winblad, B., Tuomilehto, J., Soininen, H., Nissinen, A., & Kivipelto, M. (2005). Leisure-time physical activity at midlife and the risk of dementia and Alzheimer's disease. *The Lancet Neurology*, *4*, 705–711.
- Santavouri, P., Heiskala, H., Westermarck, T., Saino, K., & Moren R. (1988). Experience over 17 years with antioxidant therapy in Spielmeier-Sjögren disease. *American Journal of Medical Genetics*, *5*, 265–274.
- Schinazi, V. R., Thrash, T., & Chebat, D. R. (2016). Spatial navigation by congenitally blind individuals. *Wiley Interdisciplinary Reviews: Cognitive Science*, *7*, 37–58.
- Singh, A. K., Phillips, F., Merabet, L. B., & Sinha, P. (2018). Why does the cortex reorganize after sensory loss? *Trends in Cognitive Sciences*, *22*, 569–582
- Singh, A., Uijtdewilligen, L., Twisk, J. W., Van Mechelen, W., & Chinapaw, M. J. (2012). Physical activity and performance at school: a systematic review of the literature including a methodological quality assessment. *Archives of Pediatrics and Adolescent Medicine*, *166*, 49–55.
- Sofi, F., Valecchi, D., Bacci, D., Abbate, R., Gensini, G. F., Casini, A., & Macchi, C. (2011). Physical activity and risk of cognitive decline: A meta-analysis of prospective studies. *Journal of Internal Medicine*, *269*, 107–117.
- Stallings, L. M. (1973). *Motor skills: Development and learning*. Boston, MA: McGraw-Hill.
- Tellevik, J. M., Storliørken, M., Martinsen, H., & Elmerskog, B. (2007). *Spesialisten i nærmiljøet* (The specialist in the local environment). Trondheim: UniPub.
- Tomporowski, P. D., Davis, C. L., Miller, P. H., & Naglieri, J. A. (2008). Exercise and children's intelligence, cognition, and academic achievement. *Educational Psychology Review*, *20*, 111–131.
- Tremblay, M. S., Colley, R. C., Saunders, T. J., Healy, G. N., & Owen, N. (2010). Physiological and health implications of a sedentary lifestyle. *Applied Physiology, Nutrition, and Metabolism*, *35*, 725–740.
- van Duyn M. A. S., McCrae T., Wingrove B. K., Henderson K. M., Boyd J. K., Kagawa-Singer M., Ramirwez, A. G., Scarinci-Searles, I., Wolff, L. S., Penalosa, T. L. & Maibach, E. W. (2007). Adapting evidence-based strategies to increase physical activity among African Americans, Hispanics, Hmong, and Native Hawaiians: A social marketing approach. *Preventing Chronic Disease: Public Health Research, Practice, and Policy*, *4*, 1–11.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.

- Wang, H. X., Xu, W., & Pei, J. J. (2012). Leisure activities, cognition and dementia. *Biochimica et Biophysica Acta (BBA)-Molecular Basis of Disease*, 1822, 482–491.
- Weil, E., Wachterman, M., McCarthy, E. P., Davis, R. B., O'day, B., Iezzoni, L. I., & Wee, C. C. (2002). Obesity among adults with disabling conditions. *Journal of the American Medical Association*, 288, 1265–1268.
- Zhu, N., Jacobs, D. R., Schreiner, P. J., Yaffe, K., Bryan, N., Launer, L. J., Whitmer, R. A., Sidney, S., Demerath, E., Bouchard, C., He, K., Reis, J., & Sternfeld, B. (2014). Cardiorespiratory fitness and cognitive function in middle age The CARDIA Study. *Neurology*, 82, 1339–1346.

Ethical Issues Related to Working with Individuals with Juvenile Neuronal Ceroid Lipofuscinosis

Stephen von Tetzchner and Finn Hesselberg

Ethical issues related to people with disabilities have received growing attention in many professions, especially issues related to life quality and rights to intervention and compensatory measures to create equal opportunities in spite of differences in abilities (Blackmer, 2000; Leahy & Szymanski, 1995). Most countries have laws that give children with disabilities equal rights to receive appropriate education. However, children and young people with disabilities are still at-risk for being left out of social and societal participation. It is an ethical prerogative for professionals to prevent or remedy such situations, and the ethical issues here are therefore about the beliefs and actions of professionals.

Most professions experience ethical dilemmas, such as lack of competence or inappropriate allocation of resources, and situations where there are contradictory ethical reasons for taking conflicting and incompatible courses of action (Howe & Boele, 2018; Knauss, 2001; Shapira-Lishchinsky, 2011). *Applied professional ethics* refers to a broad system of principles of conduct that guide and regulate the actions of people in a professional role, which are usually taught as part of the professional education (Jacobs & Hartshorne, 2003). Professionals facing ethical dilemmas have three main guideposts to help choose between right and wrong: laws, professional codes of ethics, and personal values and beliefs (Darden, 2014). The professions' national and international ethical guidelines are designed to help them in such situations (Clark, 2012; Clemente, Espinosa, & Urra, 2011; Conley, 2013; Gauthier & Petifor, 2014; Glossoff & Cottone, 2005; Leach & Leong, 2010; McNamara, 2011). However, ethics go beyond the regulations of the ethical guidelines; in fact, the spirit of the guidelines is not only to avoid doing wrong but rather to have a positive impact on the life of the student, client or patient (du Preez & Goedeke, 2013; Knapp & VandeCreek, 2004). Moreover, moral acts transcend the legal rights of the individual. Acting ethically does not merely imply following a legal rule but rather a human duty in a Kantian sense (Wood, 2007), where *respect* for the individual's dignity is added to the acknowledgement of the

law. Most important is the ability to see a person with a disability as a person instead of as an expression of an impairment. For the professional, transcending the values rather than the laws of society is the essence of advocacy.

Ethical reflection and professional practice

All educational and clinical work with children, adolescents and adults involve a certain degree of intrusion into their privacy and family life. The most fundamental ethical issue is whether assessments and interventions are sufficient to benefit the child and the family (Glosoff & Cottone, 2010). The challenges of the family and the person with juvenile neuronal ceroid lipofuscinosis (JNCL) make them vulnerable (see Chapter 24), a particular ethical responsibility therefore rests on professionals working with this group to facilitate cooperation and dialogue. Parents must be given sufficient insight into appropriate goals and methods and the children must be given enough information to understand their educational needs and social situation (Glosoff & Cottone, 2010). On a macro level (Bronfenbrenner, 1979), the cultural attitudes and attribution of worth to people with disabilities will be reflected in how the agencies are organized and collaborate (Knapp & VandeCreek, 2007). The dilemmas emerging from the education and habilitation work should be subject to ethical reflections and within a larger professional group when such a group is needed for a broader discussion of appropriate goals.

It is an ethical necessity to protect children and young persons with disabilities from incompetent and non-appropriate practices and to make sure that the measures implemented are relevant (Falvo & Parker, 2000; Glosoff & Cottone, 2010). There is a risk that the moral standards that are applied to people with disabilities are lower than for other citizens, in intervention, social interaction and other aspects of life (von Tetzchner & Jensen, 1999). The basis of such risks is not only in the person's disability but rather in the person's relationship with the professionals. However, symmetry in such relationships is difficult to achieve, particularly when the expressive means of the person with disabilities are reduced. The asymmetry implies a relationship with an uneven distribution of power, which has implications for professional practice.

Terms are the tools of ethical professional reflection and the distinction between Aristotle's terms *praxis* and *poiesis* may be useful when planning and evaluating education and habilitation work (Skjervheim, 1964). *Praxis* (or primary morality) refers to the everyday acts, acts which involve interaction with other human beings, both in private life and elsewhere. They are not valued according to the results they produce, but whether they are following the cultural standard, habit, and tradition. They concern how one conducts one's life in relation to people

who are close and toward strangers. Praxis is also a matter of honesty and of acting truthfully in real life. In praxis, the value of a person with severe disabilities, who may not be able to contribute to the common good in the community, cannot be considered less than that of people without such disabilities.

Poiesis (or secondary morality) refers to acts which have a meaning beyond themselves, which seek to achieve certain goals in the future, such as education and habilitation work. They are valued according to their intention as well as their final result. This is an important element, because the final results of education and habilitation measures are not possible to predict precisely, neither growth nor decline. The right *poiesis* acts are those that lead to the desired state and are performed in a professional sense.

For professional practice, the relationship between praxis and *poiesis* is a core issue. According to Skjervheim (1964), it is praxis which makes *poiesis* possible, and the morality of everyday acts is thus the basis for the ethical status of professional practice. The fact that *poiesis* depends on praxis implies that it is the *praxis* of the person that determines the moral status of the situation and, through reflection, the ethics and the intervention to be performed. There is no clear divide between the ethics of everyday life and professional ethics: How professionals act in their everyday life should also be reflected in their professional life. This also reflects that professionals do not approach their practice in a moral vacuum. They have a background of personal beliefs that becomes a part of their professional work and duties.

Skjervheim (1964) expresses concern that this relationship between praxis and *poiesis* is being overlooked, that *poiesis* has become primary in education. He points out that one cannot do in the technical realm what one cannot do in the ethical. *Poiesis* and technical acts must be based on praxis and ethical reflection. This is illustrated in his view on developmental psychology and upbringing: To act technically means to act on the basis of a calculation of what will happen if one does this or that. Upbringing – or parenting – belongs to the general human praxis, where the attitude behind the acts is more important than choosing the right means in the technical sense. But where the attitude is decisive, it is not primarily a question of psychology, but of ethics. According to Skjervheim (1964), goals of intervention – which belong to *poiesis* – can only be achieved if they are based on praxis. However, in discussions of ethics related to disability from a psychological and educational perspective, the *praxis* perspective may be difficult to detect. Focus is typically on the quality of the work of the professionals from a technical perspective, for example, whether teachers have selected the right goals and teaching methods or strategies. These discussions of ethics differ little from professional discussions in general. Although professional issues are basically ethical, the focus tends to be on the technical aspects, and ethical dilemmas may be hidden behind questions related to form and execution.

In a similar vein, Tranøy (1994) distinguishes between *morals* and *ethics*, where morals concern what is good and bad, and right and wrong, in everyday acts of life. Ethics comprises a more explicit, systematic and elaborated treatment of moral issues. Implicit in this distinction is the fact that ethics is related to consciousness or awareness based on reflection on moral phenomena. These phenomena are part of the living of everyday life, as well as of professional practice, and highlight the reciprocal relationship between practice and everyday life. Professional ethics are reflections on the moral implications of professional practice. In this sense ethics is a bridge between the two aspects of being a professional person: the professional side with its technology or *poiesis* on the one hand, and the everyday-life experiences or *praxis* on the other. At the same time, it mirrors the reciprocity between the life of the professional and the life of the person with disabilities, their mutual lifeworlds.

In the professional literature, ethical considerations are primarily related to the roles of the professionals but morality is in the interchange between the professional life and the supported life of the person with disabilities. The moral of professional actions is a question of whether the individual with a disability is treated as a person with dignity. For professionals, the person with a disability is neither a family member nor a person they may feel they belong to, a fact that makes a significant difference in the relationship, compared to family and other people who are close to the person with a disability. In the professional and scientific literature, people with disabilities are typically mentioned as "patients" – in a sense "objects" – although the patient role is only one of many roles in human life. The transformation from object (or patient) to subject implies seeing the individual with a disability as a person, not as a set of symptoms of a particular disease or impairment, and accepting that the person's limitations and differences still constitute a basis for a life with meaning.

When a person with JNCL is losing the ability to communicate (see Chapter 6), it influences life in more ways than just self-expression. It will change the interaction and how the person is perceived and treated, also when alternative means of communication are provided. A truly moral professional practice will depend on the acceptance of expressions of the person with a disability as authentic, even when communicative means are limited. This means that it is perceived as an actual expression of the individual's thoughts (Habermas, 1983). To acknowledge another person's communication as authentic also implies an acknowledgement of what the other person says as having value and worth taking into consideration, not only in order to achieve a particular goal, but to reach a shared understanding. Accepting a person also means acknowledging the person's inability to express meaning in a conventional manner and attribute meaning and decision to all his or her acts, also the unconventional ones. However, although it

is an act of intended acceptance, it creates a risk for attributing goals, wishes and ideas that the person does not actually have, and for erroneous interpretations of the person's behavior and intentions. Krivohlavy (1996) describes a seriously ill woman who whispered *water*, and a care worker brought a glass of water to her on the assumption that she expressed a wish to drink. However, she did not drink the water. She was asked whether she had said *water* and confirmed this. Only when the care worker went to her side and followed her gaze did he understand the meaning of the utterance: she was looking at a flower. When the water in the glass was poured into the vase of the flower, she smiled her acknowledgement. In order to understand the woman, it was necessary – as in all true communication – to take her perspective.

When people have severe cognitive and other disabilities, it may be necessary for professionals to guide and assist their choice-making, and in some situations, take decisions on behalf of the person in collaboration with the family. However, even with the best intentions professionals may take over and dominate the choices of the person. Respect for the autonomy of others and their freedom to do as they choose is a core element of ethical action (Knapp & VandeCreek, 2004; Raines & Dibble, 2011). Building the person's autonomy or self-governance therefore is a basic element of ethical professional work. A person may need help and assistance but should not be unnecessarily controlled by others, even if personal limitations may require interdependent rather than independent choice (see Chapter 16).

The understanding, respect and value that are the foundation for ethical practice also imply acknowledging that education is the normal situation for children and adolescents with JNCL. This may include the use of mainstream education as long as – but not longer than – this can provide meaningful activities and inclusion. The person should be in familiar environments to reduce the effect of dementia and declining eyesight, planning should be for a satisfying today and tomorrow's necessity, with possibilities for pursuing personal interests, goals and a good life. Yet there is very little research about education for children and adolescents with JNCL. This may reflect that their education is considered as being of little value, even if the school is a central element in their life, as it is for children and adolescents with typical development. If valued as *poiesis*, their future contribution to society may be little, and hence the value of the education because it may not lead to a profession or working life in the ordinary sense. However, when seen in the perspective of *praxis*, the education of children and young people with JNCL has its own value and equal to that of others. From this perspective, schools should provide positive challenges rather than an "easy" life with little possibility for meaning-making. In the present project, there were comments indicating that teachers and other staff found it difficult to make

appropriate plans, find the right content of the education, or giving education at all (see Chapter 11). These comments demonstrate the need for ethical reflection about human values and the good life (Barker, 2011).

Understanding the lifeworlds of people with disabilities

Professionals' understanding and attitudes toward people with disabilities will be a basis for their ethical reflection and practice. Their education and competence will influence their decisions but also less well-founded assumptions about brain training or "neuromyths" (Ansari, 2015). The professionals' understanding of an individual or group of individuals should take the whole life situation into account and not be based on a few features, such as the symptoms of a disability. All aspects of the person and the person's environment are integral to the identity and meaning-making of the person, the strength and challenges of the person, culture, history, and the societal organization and structure (Bronfenbrenner & Morris, 2006).

There are two typical ways in which a professional may develop a false understanding of the situation of persons with disabilities. Firstly, the professional may keep distance and see the situation from a narrow technical-professional perspective. The result may be an approach that focuses on instrumental achievements and less on understanding the person's sometimes idiosyncratic meaning-making. The second way is when professionals focus on their own feelings. For some, the perception of people with a disability appears to be partly based on their fear of seeing themselves in a similar situation (see Tomkiewicz, 1996). This may lead to a false feeling of being empathetic and able to evaluate what a good life is for that other person. This in turn increases the probability that the professional's own views and emotions will be attributed to persons with disabilities as if they did not have their own life. It is necessary for professionals to see both the person with a disability *and* themselves as equal partners in the relationship. Similarly, when talking about people with disabilities, politicians and others may show false empathy by saying that «all people are a little disabled or have special needs», thereby expressing a lack of acceptance of a person who is truly disabled and disregarding or minimalizing the everyday practical problems and challenges related to having a severe disability.

These possible conceptions of persons with disabilities have in common that they are not based on a true dialogue. The professional may not have listened to the person with a disability or looked for details in his or her lifeworld. Only through a broader perspective and appreciation of their everyday challenges, it

is possible for professionals to obtain an authentic impression of the situation of the disabled person. Such an insight is necessary for understanding what a meaningful life in atypical circumstances might be, and hence for planning and implementing educational and other interventions for people with JNCL or others who are losing abilities. Of importance here is the professional's realization that the learning and developmental progress of the person cannot be used to measure the quality of the work of the professional – even the best educational adaptations and interventions cannot stop the progress of symptoms in persons with JNCL.

These ethical reflections point to a wider perspective. For example, children and young persons with JNCL have different reactions to a difficult life, and anger, aggression, depression and apathy – when the person understands and manages less, and hormones rage as in puberty – should be met with *understanding*, if not with accept. Emotional and behavioral reactions need to be interpreted within the life situation of the person – a context characterized by increasing life confusion. The reactions should not be perceived as "bad" behavior and it is important to remember that the "challenge" in challenging behavior rests in those who are challenged, not in the person whose behavior elicits the challenge (Emerson & Einfeld, 2011; von Tetzchner, 2004). A similar understanding based on ethical reflection may apply to collaboration with parents. Parents of children with JNCL experience a difficult life situation, resulting in varying moods that from time to time may make positive and effective collaboration difficult. In such situations, the professional should not be aggrieved but show sympathy, understanding and acceptance – and openness if and when parents change.

Disability, ethics and human rights

Ethical reflection is a foundation of planning and implementing education and other interventions and is closely related to human rights as expressed in the UN Universal Declaration of Human Rights because human rights have underlying moral imperatives (see Gauthier & Pettifor, 2014; Nickel, 2007). In this context, reflection on ethical practice and human rights is about the values guiding the education and habilitation of people with JNCL and others with extensive needs. It is about what is best for the person with JNCL. The interventions that are selected should be satisfying today and useful for tomorrow. Childhood has a value of its own and represents a way toward adulthood.

In this perspective, a person is not just a composite of different parts but an integrated whole with continuity and change. Similar formulations may be found many places in this book. However, the demands on intervention imply choices that may give rise to dilemmas that do not have any obvious solutions. For

example, participation is a core concept in ICF (World Health Organization, 2001) and habilitation work, but may represent a dilemma when the person does not want to engage in activities which others consider necessary for a better future life. Much habilitation work is done on a very personal and private level, entering into other people's home. The following statement, based on the views of many parents in the current project (Appendix A), shows the importance of considering pro-active learning:

It was too late to start with the learning program; the cognitive decline had gone too far.

Some professionals considered it unethical to address a problem, which was not yet present. They also expressed doubts about the student's motivation to learn something he or she did not need in their current situation.

I am not sure it is fair to make plans when you know the outcome of the disease.

It feels wrong to make plans for the future when nobody knows what is going to happen.

Precautionary or pro-active teaching was also sometimes perceived as *odd*, in particular when the students were not aware of what to expect from their own diagnosis. However, parents and professionals also gave good examples of pro-active learning for students with JNCL (see Chapters 11–14). It was a matter of enthusiasm and dedication by people in the planning group, and how the teaching activities were accomplished and organized.

Participation in education that is based on the student's inability to take active part in an activity may pose a severe threat to students with JNCL and represents an ethical dilemma (Mortensen, 2006; von Tetzchner & Jensen, 1999). An Individual Education Plan (IEP) should always express the importance of education. Maintaining educational and social participation will be one of the most important requirements in IEPs for children and adolescents with JNCL. The participatory perspective should be explicitly formulated in IEPs for all students with JNCL, as well as in long-term Habilitation Plans (see Chapter 11).

Confidentiality is essential, but so is information. This is a sensitive issue with diseases like JNCL. Educational and other interventions may involve many persons and different settings. Both professionals and classmates may need to know something about the problems they see that the student with JNCL is struggling with. In addition, awareness of the possible negative consequences of not

introducing proactive or precautionary teaching makes it necessary to discuss how and when to talk about the future, and what should be told, for example in relation to the need for early learning of braille (see Chapter 14). In such a situation it is necessary to ask «what is really needed to be known by whom» (Kjønstad, 2010; Lindén & Rådeström, 2008). To solve this kind of dilemma there must be time for discussions between the family and professionals. There are no "right" answers to such questions and dilemmas, but it is important to find a practical consensus.

In the same vein is the question of keeping the disease secret from the individual with JNCL and people in the environment. This has been an ongoing discussion among professionals with little parental participation (Elmerskog & Fosse, 2012). The only acceptable solution to this dilemma is simple: it is the parents who have to make such a decision on behalf of themselves and their child, without their decision being questioned or reviewed by professionals. This is illustrated in the present project, where two pairs of parents had made different decisions and both expressed that their decision was a successful one (see Chapter 25). Each family has built their own context and decisions are made within this context. However, this does not mean that professionals should avoid such delicate themes but should contribute with objective information and their experiences when asked, expressing a clear attitude that such issues are for each family to decide and cannot be divided into right or wrong solutions.

At an organizational level, it is important to obtain a consensus between the family and the services regarding education and intervention for the person, especially what the family should and can expect of the services. When decisions to be made involve professional issues and values, it is also an ethical issue how the decisions will affect other services and persons, and who is entitled to make the decisions. Sometimes it may seem to be solely a professional decision but in reality, it is more of a moral decision; it is about values and attitudes, such as when there is a choice between a specialized institution relatively far away from family and peers or a more mainstream solution nearby the home (Hesselberg & von Tetzchner, 2016).

Ideal and pragmatic services

Ideal educational and other intervention measures are based on professional sensitivity to the family and the student (Hesselberg, 2017). For example, the amount of professional follow-up needed and the form this should have, will be one of the first joint decisions of the family and the professionals. The aim is to establish an understanding where parents and professionals share expectations to the collaboration and can raise the issue if these expectations are not met. In

addition, knowledge and competence of teachers and other staff should be sufficient and predictable. There must be local knowledge as well as knowledge about the disease and what to do. Poiesis implies being trustworthy and committed to the ethical stance *and* to strategies that are proven to be useful – in this way, the evidence base is a theoretical and empirical foundation of both practical work and ethical reflection. Professionals should have authority, which means that their knowledge should be substantial and presented in a believable way. Importantly, they must know the limits of their own knowledge and where to get help with things they do not know well enough or are unable to manage themselves. This implies knowing the limits of their responsibility, what decisions they can and cannot take. The ethical stance requires routines for making joint decisions about education and special education, and about providing challenges and care (Raines & Dibble, 2011).

The need for ethical reflection may be most evident in working with plans for the future. The Habilitation Plan and the Individual Educational Plan (see Chapter 11) need ethical reflection to ensure that plans are based on praxis and poiesis. Habilitation Plans involve personal and private matters, results from assessments, and decisions affecting the whole family. Professionals must acknowledge the variation between individuals with JNCL, for example related to age, problems and circumstances of life, support and counseling. Ethical reflection is needed for making plans for education and intervention, but plan-making in itself also contributes to raising awareness of the need for such reflection. Ideal and pragmatic services seek to establish an *alliance* between the family and the professions, with collaboration, engagement and insight into the ethical dilemmas that may challenge the overall plan and sub plans (see Chapter 11).

Concluding remarks

Working with children and young people with JNCL and their families may imply many challenges and ethical dilemmas. An ethical practice implies searching for the balance between the focus on the life today and the needs of tomorrow, and between positive learning challenges and social participation. Ethical dilemmas are always difficult, this is why they are dilemmas, but the answer is not neglect but ethical reflection. Morals, even if applied to the individual, are basically social, reflecting the norms and values of the society. Moral development is not conceivable outside a community of some kind. It is, however, not enough to follow the habits and traditions of the society, it is these that the individual has to take a stand on and transcend. It is possible to reflect on ethics in solitude, but ethics can only be cultivated in a social context and through discourse. To ensure that

interventions have an ethical foundation, discourse should be facilitated between various professionals, family, and other significant persons in the life of the person with JNCL. Communication with the person, even if severely and profoundly impaired, should also be part of this process.

Reflecting on ethical dilemmas is an important element of working with education and habilitation for children and young people with JNCL. However, ethical reflection is not only about the basic existential questions but also about a fundamental respect for the individuals with JNCL and their families, as well as for those who are doing the everyday interventions, from assistants to specialized professionals. One's ethical stance and attention to reflection and reflectivity are equally important for the responsibilities of determining practical adaptations of everyday activities and of participating in discussions about life choices. Both responsibilities require knowledge, understanding and sensitivity to be addressed in a systematic and good way.

References

- Ansari, D. (2015). Mind, brain, and education: A discussion of practical, conceptual, and ethical issues. In J. Clausen & N. Levy (Eds.) *Handbook of neuroethics* (pp. 1703–1719). Dordrecht, NL: Springer.
- Barker, P. (2011). Ethics – In search of the good life. In P. Barker (Ed.), *Mental health ethics* (pp. 5–30). Abingdon, UK: Routledge.
- Blackmer, J. (2000). Ethical issues in rehabilitation medicine. *Scandinavian Journal of Rehabilitation Medicine*, 32, 51–55.
- Bronfenbrenner, U. (1979). *The ecology of human development*. Cambridge, MA: Harvard University Press.
- Bronfenbrenner, U., & Morris, P. A. (2006). The bioecological model of human development. In W. Damon & R. M. Lerner (Eds.), *Handbook of child psychology, Sixth edition, Volume 1: Theoretical models of human development* (pp. 793–828). New York, NY: John Wiley.
- Clark, C. M. (2012). Individual moralities and institutional ethics: Implications for the Canadian Code of Ethics for Psychologists. *Psychotherapy and Politics International*, 10, 187–204.
- Clemente, M., Espinosa, P., & Urra, J. (2011). Ethical issues in psychologists' professional practice: Agreement over problematic professional behaviors among Spanish psychologists. *Ethics and Behavior*, 21, 13–34.
- Darden, E. C. (2014). Ethics at school: Let your conscience be your guide. *Phi Delta Kappan*, 95, 70–71.
- Darr, K. (1991). *Ethics in health services management*. London, UK: Health Professions Press.
- du Preez, E., & Goedeke, S. (2013). Second order ethical decision-making in counselling psychology: Theory, practice and process. *New Zealand Journal of Psychology*, 42, 44–49.
- Elmerskog, B., & Fosse, P. (Eds.) (2012). *Lære for livet. En innføring i pedagogikk og læring ved juvenil neuronal ceroid lipofuscinosose (JNCL)* (Learning for life. Introduction to teaching and learning in JNCL). Trondheim, Norway: Akademika.

- Emerson, E., & Einfeld, S. L. (2011). *Challenging behaviour, Third edition*. Cambridge, UK: Cambridge University Press.
- Falvo, D. R., & Parker, R. M. (2000). Ethics in rehabilitation education and research. *Rehabilitation Counseling Bulletin*, 43, 197–202.
- Gauthier, J., Pettifor, J., & Ferrero, A. (2010). The universal declaration of ethical principles for psychologists: A culture sensitive model for creating and reviewing a code of ethics. *Ethics and Behavior*, 20, 179–196.
- Glosoff, H. L., & Cottone, R. R. (2010). Rehabilitation counselor education and the new code of ethics. *Rehabilitation Counseling Bulletin*, 53, 243–248.
- Habermas, J. (1983). *Moralbewusstsein und kommunikatives Handeln*. Frankfurt, Germany: Suhrkamp Verlag.
- Hesselberg, F. (2017). Fagideologiske og etiske utfordringer i den pedagogisk-psykologiske tjenesten (Professional and ethical challenges in the pedagogical-psychological agency). *Psykologi i kommunen*, 52, 21–33.
- Hesselberg, F., & von Tetzchner, S. (2016). *Pedagogisk-psykologisk arbeid* (Pedagogical-psychological work). Oslo, Norway: Gyldendal Akademisk.
- Howe, K. R., & Boele, A. E. (2018). *The ethics of special education, Second edition*. New York: Teachers College Press.
- Jacob, S., Decker, D. M., & Lugg, E. T. (2016). *Ethics and law for school psychologists*. Hoboken, NJ: John Wiley.
- Kjønstad A. (2009). *Taushetsplikt om barn* (Confidentiality related to children). Oslo, Norway: Kommuneforlaget
- Knapp, S., & VandeCreek, L. (2004). A principle-based analysis of the 2002 American Psychological Association's ethics code. *Psychotherapy: Theory, Research, Practice, Training*, 41, 247–254.
- Knapp, S., & VandeCreek, L. (2007). When values of different cultures conflict: Ethical decision making in a multicultural context. *Professional Psychology: Research and Practice*, 38, 660–666.
- Knauss, L. K. (2001). Ethical issues in psychological assessment in school settings. *Journal of Personality Assessment*, 77, 231–241.
- Krivohlavy, J. (1996). *Existential dialogue in contemporary psychotherapy*. Lecture presented at the University of Oslo, Norway, October 9th 1996.
- Leach, M. M., & Leong, F. T. L. (2010). International dimensions of psychological ethics. *Ethics and Behavior*, 20, 175–178.
- Leahy, M. J., & Szymanski, E. M. (1995). Rehabilitation counselling: Evolution and current status. *Journal of Counselling and Development*, 74, 163–166.
- Lindén, E., & Rådeström, J. (2008). Ethical dilemmas among psychologists in Sweden and South Africa. Professional thesis, University of Linköping, Sweden.
- McNamara, K. M. (2011). Ethical considerations in the practice of school psychology. In M. A. Bray & T. J. Kehle (Eds.), *The Oxford handbook of school psychology* (pp. 762–773). Oxford, UK: Oxford University Press.
- Mortensen, E. (2006). Ethical perspectives on working with students who have Batten disease. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 71) Örebro, Sweden: Swedish Institute for Special Needs Education.
- Nickel, J. W. (2007). *Making sense of human rights, Second edition*. Oxford, UK: Blackwell.
- Raines, J. C., & Dibble, N. T. (2011). *Ethical decision making in school mental health*. Oxford, UK: Oxford University Press.

- Shapira-Lishchinsky, O. (2011). Teachers' critical incidents: Ethical dilemmas in teaching practice. *Teaching and Teacher Education*, 27, 648–656.
- Skjervheim, H. (1964). *Vitskapen om mennesket og den filosofiske refleksjon* (The science of man and the philosophical reflection). Oslo, Norway: Tanum.
- Tomkiewicz, S. (1996). *Ethics and disability*. Paper presented at the Ethical Challenges in the Support of People with Severe Disability Conference, November 1996, Lisbon, Portugal.
- Tranøy, K. E. (1994). *Medisinsk etikk i vår tid, Annen utgave* (Medical ethics in our time, Second edition). Søreidgrend, Norway: Sigma.
- United Nations (1983). *World programme for actions concerning disabled persons*. New York: United Nations.
- von Tetzchner, S. (2004). Early intervention and prevention of challenging behaviour in children with learning disabilities. *Perspectives in Education*, 22, 85–100.
- von Tetzchner, S., & Jensen, K. (1999). Interacting with people who have severe communication problems: Ethical considerations. *International Journal of Disability, Development and Education*, 46, 453–462.
- Wood, A. W. (2007). *Kantian ethics*. Cambridge University Press.
- World Health Organization (2001). *International Classification of Functioning, Disability and Health: ICF*. Geneva: World Health Organization.

9

Organization of Education and Counseling

Evelyn de Lorent, Cari Mannion, Marja-Leena Forssas, Tarja Eskonen, Anne Hagedorn Hamann, Susan Fugger, Merete Staureby, Bengt Elmerskog and Finn Hesselberg

The organization of the educational system is a foundation for the education of all students. However, it is especially important for students who have a disability because they need educational structures and functions that are tailored to their needs and possibilities. While most children will develop and learn under varied conditions, children with disabilities depend on a narrower set of structures and functions in order to develop and learn optimally (von Tetzchner, 2019). The organization also varies in geography and size, culture and history, and the national traditions and ideas about education.

The present chapter presents elements from the educational systems in five countries (Germany, Scotland, Finland, Denmark and Norway) that are relevant for the education of children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL), including some case stories. In all the countries, support for the family and collaboration with parents are important parts of the work (see also Chapter 24). In the present project (Appendix A), the percentage who attended mainstream school was gradually reduced with age. More than 80 percent of the participants with JNCL had attended a mainstream school in grade 1 and 2. About half had attended a mainstream school in grade 5, and in the higher grades (8+), more than 80 percent had attended a special unit or school.

Germany: Educating children and adolescents with JNCL in Hamburg

Germany has a size of 357,170 square kilometers and a population of 81 million (2014). It is a federation of 16 states and Berlin is the capital with 3.5 million inhabitants. Hamburg is a state and the second largest city with a population of 1.8 million. It is located in the north of the country.

The Education Centre for the Blind and Partially Sighted (BZBS) in Hamburg has provided education for children and adolescents who have JNCL for several decades. A JNCL working group was established many years ago to organize and monitor the education of this group of students. The work presented here is related to students starting school, transitions (changing class or school and leaving school) and collaboration with parents.

Starting education

The course of the disease varies significantly, and the school experiences and careers are also quite diverse for students with JNCL. Most of the students with JNCL who have attended BZBS began their education in a mainstream school. The eye condition usually became noticeable during their first two years at school (see Chapter 4). In this challenging situation, the dilemma arises as to where the child will receive the best possible education. The options were either that the child stayed in the mainstream school with special educational support or moved to the Education Centre for the Blind and Partially Sighted.

The decision about whether it is best for the child to move to a special school should be carefully reviewed and it is essential that it is made on the basis of a close and trusting relationship between counselors, special education teachers, the ordinary teachers and the parents. Experience in Germany indicates that an immediate change of school, especially and preferably in the early years (primary school), is most effective. Depending on the course of the disease and the child's condition, a further decision is made about placement in BZBS: whether the child attends an ordinary class or a special needs class (for students with multiple disabilities). Both may be a good choice, depending on the needs of the individual child.

The parents initially find themselves in a seemingly inexplicable, threatening and extremely stressful and challenging situation. Embroiled in the difficulty of coming to terms with the diagnosis of visual impairment, they encounter new changes which at first seem to be related to the emerging blindness. Parents live through the fear and unhappiness of their child and find themselves in a state of exceptional stress.

The immediate changes experienced by the child at this time make him or her increasingly anxious. The child tries to cope, compensate or even hide what is happening. At the same time, the people around the child are also experiencing insecurity, uncertainty and perplexity, as well as feelings of guilt. The changes in the child's daily life, such as starting school or changing school, and an odyssey of visits to hospitals and other institutions, imply numerous stressful and frightening situations.

It usually does not take a long time from the initial observations of the child's deteriorating vision until changes in behavior and loss of motor functions become noticeable. Learning challenges typical of children with JNCL are identified by the special education teachers and the final diagnosis is soon made. The diagnosis of JNCL is a huge shock to parents and places them under enormous pressure. It is difficult for them to come to terms with the situation. They need to consider how to support their child and at the same time look after their other children, as well as deciding how and when to explain the situation to the child and the siblings. The issues of how to inform others and speak about the illness need consideration and are challenging to all parties involved. When a child is transferred to a school for the blind, it is important that the school is aware of the personal situation of the child with JNCL. Sensitivity to the child's condition and an understanding of the child's situation are important, including the home situation and the atmosphere in the home and other parts of the immediate environment.

To this end, the school offers many opportunities for dialogue with the parents, where the support options and forms of help available are outlined without exerting any pressure. The child's lessons are modified to address the changes which may not yet have substantially affected the teaching and learning process.

The speed and extent of the changes in behavior, cognitive decline, reduced motor functions, speech problems and onset of seizures vary, and these may appear together or in succession (see Chapters 3–7). However, irrespective of the course of the disease, the emergent declines have serious effects on the child with JNCL and impact every area of the child's life – family, social life and school.

The child or young person will often be keenly aware of these changes and will ask «What is happening to me?» and then «Why?» They may become extremely distressed, show fear and despair, and at the same time go through periods of denial and suppression, not wanting to acknowledge what is happening.

As the cognitive functioning of the child or young person is gradually affected they become less capable of a realistic appraisal of their own situation and the options to manage these changes will be fewer. Parallel to the changing reactive behavior, the disease also may imply changes in behavior and character that bring additional problems to the group situations. Young persons with JNCL tend to develop an insistence and reliance on specific routines and patterns of behavior, which can be very personal and idiosyncratic. It is not always possible to accommodate these behaviors in the class, and this may lead to stress and conflict in the classroom.

It is necessary to look at each child or young person individually and constantly review the organization and teaching content to determine where they can be best supported. A move to a special needs class should always be

thoroughly discussed with all parties involved (parents, current and new teachers) and the transfer should be carefully prepared and implemented.

Moving to a special needs class

Many students with JNCL are becoming sensitive to change and moving to another class should be considered only when absolutely necessary. It is important to reduce the stress of having to adapt to new conditions, places and people. A gradual phasing-in of such changes seems to be the best way to reduce the possible strains. The student will be introduced to the new class in the course of several visits and the duration of the visits will gradually be increased. When entering the new student group, the student with JNCL will gently be introduced to the new peers and to new but interesting media and materials, such as the computer program *Sarepta* (see Chapter 19).

The student may observe a discrepancy between his or her own ability level and that of the new classmates, and this could prove problematic as the level of the class might be clearly lower. However, this can also have a positive and motivating effect, as the student with JNCL may feel less pressure to perform or feel superior. At this time, the student may have considerable skills and competence that can be used in the classroom. When it is possible, the student can continue having lessons in a favorite school subject (such as sport or music) in the former class and thus maintain his or her existing social relations.

Generally, the student with JNCL will become less perceptive of the situation and less aware of the changes and declines. Teachers must be attentive to signs or behaviors in the students, in order to respond appropriately to their changing needs and ensure that the right help and support are provided in an atmosphere that is peaceful, secure and comforting.

Cooperation between school and home

Naturally, the teachers, special educators and other members of the school staff cooperate closely with parents. Sufficient time needs to be allocated to collaboration with parents as it is evident that the education and support provided to the student with JNCL in school are greatly determined by the quality of the cooperation and interaction with parents.

How the communication between school and parents is conducted and how important facts are relayed will depend on several factors. It is the school's experience that the relationship that is formed between school and parents is greatly influenced by the personalities involved. Some parents are very approachable and desire regular communication, while others may prefer more sporadic contact.

The location of personal meetings should, at first, be decided by the parents according to their wishes. Inviting parents to the school, at least during the first period, may trigger uncertainty and anxiety. One well-tried approach is the «round-table talk», where a psychologist invites the parents to a meeting to discuss specific issues with family members and class teachers, and then moderates the meeting. The team members are thus relieved of the problem of how to manage the discussion and what they sometimes refer to as an «emotional chaos», allowing them to focus on the issues discussed in the meeting.

Meetings can involve several parents and teachers, which facilitates the exchange of information and experience. This may lead to a continuous exchange of updated information and a network directory, an exchange of day-to-day experiences, expectations and feasible outcomes. Importantly, such meetings offer time and place for expressing anger and grief, as well as for sharing the fun and enjoyment that can be encountered in daily classroom situations.

It is not a question of who has the highest level of professionalism with respect to the care of and relationship with the student. The parents are always

Table 9.1 The school’s statements for guiding an ethical practice

The following core statements form the ethical basis for our pedagogical and therapeutic mission. They express our commitment to an authentic, professional approach. The code commits us to a serious and honest management of the individual characteristics and condition of every child with JNCL, both in our day-to-day actions and in every discussion and consultation. Implicit to this code is open and honest communication between all staff in this institute and the parents, based on trust, respect and acceptance.
Every child has the right to a fruitful life and the chance to develop his/her personality as fully as possible.
The rights and needs of a child with JNCL are of no lesser value and are equal to the rights and needs of other pupils.
We ensure that the dignity of the child is observed and tolerate no belittlement or disrespect.
Our mission is to make every day as fruitful and meaningful as possible.
We encourage and foster the child to help it develop its full potential within his or her capabilities.
We accompany and support the child through the process of change and deterioration.
We are committed to ensuring that the child is part of the community and group experience for as long and as far as possible.
We protect the child from the risk of becoming lonely and isolated.
The child has the right to know the truth. Working closely with the parents we gradually reveal the truth of the situation.
While we will never lie to a child, we will also never deny him or her any hope.

the center of all decisions. Together with the school, they are responsible for achieving agreement concerning responsibility and authority. An essential part of such agreements, or alliances (see Chapter 10), is consultation on informing all the persons and groups involved with the child about JNCL. Through long experience with working with children and adolescents with JNCL, BZBS has accumulated basic know-how which has been formulated in writing as an internal ethical consensus or code of conduct, which is binding for all educational and other personnel. These ethical principles are the basis for a code which governs professional behavior and treatment of the students with JNCL and their fellow students, cooperation with families, cooperation within the institution and with other schools, specialized agencies and cooperating institutions and partners (see Table 9.1). The application of this internal school code to other agreements, which define mutual commitments and responsibilities, has proven useful. The statements and how they are applied on a day-to-day basis in work are constantly reviewed and scrutinized. The work of BZBS is carried out within the context of a continuous exchange and mutual support with other specialist disciplines (see also Chapter 8).

Transitions and leaving school

Schooling generally finishes at the end of the statutory school-leaving age, although sometimes an application might be made to end it earlier or beyond the maximum statutory age limit, depending on the development of the child, his or her life situation, and the family's circumstances, all of which are important factors in this process.

A place in an institution after leaving school will usually be made available at short notice to persons with JNCL. This offers new perspectives and opportunities for the individual, as the transition from school to further training is rare for persons with this condition. In such cases it is necessary to prioritize the preparation and encouragement of the person and facilitate a smooth and unbureaucratic transfer (this could entail leaving school mid-term or before the end of statutory school education).

The following principle applies: schooling should be organized as flexibly as possible. Any decision about subsequent suitable vocational training should be made on an individual basis taking the person's needs and abilities into account. Depending on the individual, the most likely options are a special day school or training in a workshop for people with disabilities. Students participate in various vocational assignments during their last three years at school, and thereby individual interests and abilities are identified, nurtured and developed with the aim of finding appropriate workplaces. Other factors are also considered, including the living and care situations, the situation of care givers and support staff and opportunities for

retreat and relaxation. During the vocational assignments members of the staff of the Center (BZBS) talk to staff from the potential workplace about JNCL, with the aim of facilitating a job offer. It is important to ensure continuity and as far as possible avoid future changes of workplace, group and support persons.

If a suitable future workplace or occupation is found, the trainer is offered the opportunity to meet and observe the student in the classroom in order to gain a better understanding of the future trainee in his or her familiar and adapted surroundings. Similarly, the Centre offers to send the class teacher to the new training place to facilitate the transition and provide support and advice at the early stage. An additional period of vocational experience at the future job or workplace while still being a student at the school has also proven useful in helping the student to prepare for the future life.

The entire process of seeking sheltered employment takes place in collaboration with the parents. The issues that need to be addressed and the specific approach are decided very much on an individual basis, asking questions like: How much support do the parents want? Will vocational activities be combined with a residence? Does the workplace need to be close to the person's residence?

Our experience over the years indicates that the transition of a student into employment is a challenging time for parents of children with JNCL. Following a fairly long period of relative peace and stability during the school years, parents again need to make important decisions for the future. They are again confronted with the degenerative nature of their child's disease and this will shape their actions and decision process.

Educational concepts and aspects of the schooling and support of students with JNCL

The review of educational work with children and adolescents with JNCL by the working group of the Center was presented in 2010, describing four core elements and principles: a) Tasks and objectives, b) teaching content, c) equipment and materials, and d) approach and methods. The first three of these elements are presented below.

Tasks and objectives

The staff of BZBS accompanies and supports children on their educational journey and ensures their dignity. Core principles of BZBS are *attentiveness*, *support* and *sincerity*. These constitute the heart of BZBS's work. BZBS has undertaken an obligation to cater for each student's educational needs and not make premature assumptions about the students' educational possibilities and limitations. BZBS

try to attain the right balance between educational challenge, encouragement and support. The staff at BZBS should have insights and perspectives related to normality, typical development and atypical development based on an understanding of disabilities and disorders. BZBS should acknowledge and understand that declines and regression are to be expected in children with JNCL. This acceptance should not exclude the possibility of providing engagement and opportunities to the students, enable their participation in society, or making every day enjoyable and meaningful. This understanding will guide the staff of BZBS when they are planning and implementing educational programs. The programs should be specific and support the following steps:

- BZBS will support the student's development by encouraging them to acquire the highest possible knowledge and skills.
- BZBS will help the students to retain and maintain acquired competence and skills as long as it is possible.
- BZBS will seek to ensure the maintenance of the students' best possible physical and mental state and health.
- BZBS will support the students' reminiscence and the maintenance of memories of special importance to them.

Teaching content

Education for students with JNCL is based on a profound contradiction between development and regression. This dichotomy is however less pronounced today, due to the diversity of learning options and attention to individual needs resulting in individualized education programs.

The students should remain in inclusive settings as long as possible, that is, within their peer group, and receive the same offers and activities as others. However, the overall aim is to promote the best possible learning, development and maintenance of skills and capabilities.

An Individualized Education Plan (IEP) will be needed when the condition of the student with JNCL deteriorates. The mainstream curriculum will at some point not meet the student's special needs. Catering for the individual needs specified in an IEP requires a broad understanding of the student's life and his or her interests and preferences (see Chapter 11). However, the educational content is not determined solely by the student's interests and preferences for subjects and themes. Education must also address goals and learning areas selected by objective means, such as maintaining independence, active participation and social relations.

Group learning and activities become easier to achieve if the special interests and skills of the student with JNCL can be integrated into the activities of the

class. As the disease progresses, the student may retreat into strong personal interests, which may function as escapes from the everyday reality. These escapes or behaviors may dominate the student's life. Sometimes the student's personal interests are so special and different from those of others that integrating them in the class activities becomes impossible. There may be situations when students should be encouraged to discover and develop interests to be used as escapes for meeting difficult realities in life. However, this may be difficult to achieve. Even the best plans may not result in achievements, as illustrated in the example below.

The beautifully designed furnished doll's house did not prove to be as successful as expected. The girl with JNCL, who we assumed would be eager to play with the house and the dolls, simply found the house to be stupid.

A varied musical curriculum is important (see Chapters 17 and 18). Other areas are role play, theatre, games, working with materials, sounds, dance and movement (see Chapters 12 and 21). These areas can be used to support feelings, expressions and communication.

Program content also includes sport, swimming, activities of daily living, physiotherapy, occupational therapy, mobility and orientation. In early phases of the disease, these activities will be used to support further learning and development, and in the later phases to maintain skills and capabilities. Many of these areas will also support participation in social settings and activities and will help students to cope with everyday challenges.

Sitting in a cozy corner together, cuddling, talking, changing positions, movement, enjoying going outdoors and fresh air are simple (yet valuable) experiences that can be easily achieved at the school grounds, on shopping trips, walks, excursions and the like.

The students may need psychological support to cope with their own situation and attain emotional stability. It is vital that the class has an atmosphere of openness, trust, safety and comfort in which a student can speak about feelings without anxiety and fear. Being open about feelings is positive for everyone, students as well as teachers, and on equal terms. This is illustrated in this example:

Students with JNCL can use a memory bank where they can store ideas, thoughts and reminiscences. The boxes will be filled with different thoughts and memories, and they can be emptied and refilled again, dependent on the student's needs for reflection and storing memories.

Explaining bodily changes for students with JNCL requires skills and strategies. It is important that the students do not feel isolated and alone when experiencing changes and declines. There are children's books and CDs that effectively address fear and anxiety, limitations, loss and separation, death and dying. This is illustrated in this example.

The book Seelenvogel (Snunit & Golomb, 1991, 1999) contains excellent images that illustrate different human qualities, concerns and aspects of life. Through role play and cuddling we used the themes of the books to make a reality play. During the session, a girl with JNCL was able to put aside her inhibitions and express freely about her anger with having this disease.

Material and equipment

It is not always possible to know what material and equipment there might be a need for as the interests of the students are constantly changing. It is a good idea to provide teachers with a collection of much-loved and tried materials and accessories. For example, unconventional materials can be used for creative work:

One boy had a special love of shoes of every kind, so the school janitor built a shoe cupboard for the whole class. The boy who loved shoes was able to share his interest with the class and the shoes became a source of discussions.

Memorable and favorite items may be stored for the future, even if the student at present is not showing particular interest in the object. It is useful to compile a collection of some kind – including unconventional objects – for every child. The «treasure chest» is a much loved and extensively used place to store memorable objects, loved items, and mementos. Some children have a treasure chest at school and at home. It may contain old toys and CDs (it is advisable to make copies!). The possessions and objects can be used for refreshing or reviving memories in the future. Students seem to like objects that can produce sounds, and sound recordings of their own voice is often a success. Sarepta is a software program that offers diverse opportunities for work and play. It can be customized to the individual and the changing needs and abilities of the person (see Chapter 19).

Scotland: Aspects of the Scottish Education System

Scotland is part of The United Kingdom. Scotland has a size of 78,282 square kilometers and a population of 5,4 million (2017). The capital is Edinburgh with a population of 450,000.

In Scotland, provision has been made by the Scottish Government through the Education Act (2000) which stipulates that individuals with additional support needs, such as students with JNCL, should be able, as a right, to attend their local mainstream school. Further the Education Act (2004) was introduced in order to provide explicit and legally binding support for children and young people identified as needing additional support within the school setting. Both Acts grant children and their parents important rights which affect education. The 2004 Act was amended by the Education (Additional Support for Learning Scotland) Act 2009 to include support from other professionals, to ensure that young people are provided with all the necessary support to help them work towards achieving their full potential within a mainstream school.

Under the Education Act 2009 (Additional Support for Learning Scotland), parents and/or anyone working with a child who concludes that the child has additional support needs, can request that the local education authority investigates to establish if this is indeed the case. If so, the education authority is legally required to make "adequate and efficient provision" to meet the identified additional support needs of the child. This process is underpinned by another framework initiated by the Scottish Government – "Getting it right for every child" (GIRFEC), which puts the child at the center of any identified initiative or plan involving other professionals. Together with "The Curriculum for Excellence", GIRFEC aims to support and improve outcomes for all children who have been identified as having additional support needs. These policies, however, at present, offer no template for provision within a mainstream setting for young people with JNCL. Therefore, the Royal Blind School is just one model of possible and potential provision as educational authorities could assert that these individuals' needs could be met in their own learning support provision within a mainstream setting.

After a process of assessment, if a child or young person is identified as having complex or multiple additional support needs, a Coordinated Support Plan (CSP) is compiled to ensure that the support for learning can be coordinated across a range of agencies. The CSP is a legal document and an educational psychologist of the education authority is responsible for monitoring, reviewing the plan, and ensuring that all set targets are being met annually.

Depending on the financial resources of individual local authorities within Scotland, the path to securing a placement in a school of choice by parents and others is not always guaranteed. In some situations, if parents are determined to secure a place of their own choice, rather than that of the local educational authority, then a tribunal takes place, which can be an arduous and demanding process, with no guarantee of the desired outcomes for the parents.

The Royal Blind School in Edinburgh is a specialist center of provision of education for young people with visual impairment. Local education authorities provide referrals and ensure funding for day or residential students with a range of additional needs. Since there is a wide range of expertise on site, young people have access to experienced staff trained in supporting young people's individual needs, including qualified teachers of the visually impaired, trained support staff, occupational therapists, physiotherapists, speech and language therapists, and habilitation and orientation specialists. There is also access to the latest assistive technology, small teaching groups, and specially tailored programs of learning through the implementation of Individual Education Plans (IEPs). The Royal Blind School also provides outreach support to other teachers and support workers throughout Scotland through its "Learning Hub", via for example short online video programs and training days in school. Staff in school continues to be involved in provision for students with JNCL and work through dedicated computer application technology and in-service training sessions within school, outside conferences, both as participants and facilitators.

Presently in Scotland, young people deemed to have an additional learning need, such as persons with JNCL, do have a choice of provision. However, the options may not always directly meet the needs of the young person. For some individuals an inclusion unit may provide the necessary support whilst for others a specialist school placement may be more appropriate.

Case story: Residential school placement in Scotland

Ideally, children would attend their local school and engage and participate in their local community. With children who have additional support needs, and specifically children with JNCL, this is not always possible. Parents want their child's needs met in an environment which supports their learning and wellbeing. It is not an easy decision to opt for a residential placement. And even when the decision has been made, funding is not always easy to access. The Royal Blind School offers weekly and part week residential placements. All residential care workers are qualified, and the service is regulated by the Care Inspectorate. Residential care workers, education staff, therapists and habilitation specialists are offered regular in-service training on

JNCL. They are all encouraged to promote excellent collaborative practice and to share innovation in their approach to the individual pupil.

This case study is to highlight for families and professionals the role of residential staff in supporting children and young persons with JNCL and their families.

Adele joined the School after it had been noted, at her local school, that there had been a significant deterioration in her vision. She was diagnosed with JNCL shortly after this. In discussion with the parents, the local education authority and the school, a weekly placement was decided as the best option for Adele. Factors that were emphasized included home circumstances, geographical location of the family, access to expert staff with knowledge and experience of JNCL. The parents visited the school and met with senior managers, including the head of care and other staff who would be working directly with their daughter. Adele was allocated a key worker who was the main contact point for the parents regarding all residential matters and a teacher to liaise regarding educational matters. The key worker and teacher met regularly to pass on relevant information and ensure that communication with the home was effective.

The key worker would attend team meetings and ensure that Adele's daily observation file was up-to-date, pass on information to relevant staff and write monthly reports. The file would contain a detailed Care Plan and IEP, daily observations, monthly reports, notes from liaison meetings, medical information, information from other professionals, updates on JNCL from in-service sessions, BDFA and conferences attended by one of the deputy head teachers. The documents would detail, for example, the need for a smaller, quieter living skills area for Adele to allow her to maintain independent living skills as long as possible, Adele's interests and aptitudes, and strategies for dealing with mood swings. The staff used a communication diary and, in accordance with the wishes of the parents, telephoned regularly, daily if requested. Adele's mother was supported by a close relative and later on by a family friend, both of whom kept in regular contact with the school. Her mother was able, if she wished, to meet with the staff any Monday or Friday by coming to school with Adele's transport. Though welcome to come at other times, transport was not easy and time consuming. Parents are also welcome to come to class to see strategies in practice.

Like all pupils at the school, Adele had reviews at least once a session and more often at times of transition. These were attended by school staff, local authority professionals and the family. Adele attended some of her reviews, being prepared and supported by her key worker and teacher.

As she entered her teenage years, Adele began to have quite dramatic mood swings. These had, at times, a significant impact on her attitude and behavior. She could be very stubborn regarding personal care and angry at her peers, not wanting to engage with them and staff. This was a pattern that was happening at home at weekends and her mother found this very challenging. The staff supported Adele by allowing her time and a quiet space. Her parent felt supported with strategies that had been tried successfully at school. The residential staff were responsible for the residents in their care from around 4 pm until 9 am the following day. This is much longer than with any other group of staff. Of course, strategies that worked at school did not always work at home, and vice versa, but the communication with the parents was vital to ensure the best possible support for Adele. The regular meetings with the educational staff also ensured that new ideas could be discussed and taken forward with time allocated to evaluate suitability and effectiveness of any adopted strategy. Adele's teacher and key worker collaborated with the family to build a memory book. This was a precious document to Adele and her family, as well as being of great use to new staff and for Adele's transition post-school. Over time Adele was also subject to hallucinations and would become extremely agitated in the evening. As her communication skills deteriorated, life became very challenging for her and, naturally, for her mother and close family.

The residential staff and especially Adele's key worker observed to identify what worked well for Adele. She liked familiar people around her when she was distressed. She loved music and that continued to be an area she connected with throughout her time at school. Adele enjoyed having her nails manicured and the company of younger peers in the residential area where she was living. Animals were of great interest to her. Any changes to her routine had to be gradual and by maintaining her interest Adele could be supported by staff to try new things or meet a new classmate or member of staff. This stimulation was seen to have a beneficial effect on her determination to keep trying. Throughout her teenage years, the staff supported Adele with the technology she used in school and encouraged her to practice her braille skills and maintain her social life. Key to Adele was music; even when her speech became unclear and she stuttered, she was able to sing clearly. She also enjoyed certain stories that she was happy to hear time and time again.

The bond that was built between Adele and the residential staff, the parents, the teacher and the support staff lasted as the staff continued to visit Adele after she had left school.

Finland: Valteri Centre for Learning and Consulting

Finland has a size of 338,430 square kilometers and a population of 5.5 million (2015). Helsinki is the capital with 621.000 inhabitants.

Valteri Centre for Learning and Consulting is a national center operating under the auspices of the Finnish National Board of Education (FNBE). FNBE is subordinate to the Ministry of Education and Culture and its organization and functions are set in the legislation. Valteri consists of six units in different parts of Finland: Mikael, Mäntykangas, Ruskis, Onerva, Skilla and Tervaväylä. According to the Law of Education, Valteri has the nationwide responsibility to administer and develop consultation and support related to inclusive education in municipalities and arrange education and rehabilitation for some children with special needs at each Valteri unit.

Valteri professionals, such as consulting teachers, class teachers and rehabilitation professionals, have considerable competence and experience related to autism spectrum disorders, neuropsychiatric disorders, disorders of language and communication, hearing, vision, mobility and motor functions, neurological diseases or other chronic conditions, and multiple needs. Valteri staff has extensive multi-disciplinary cooperation with schools, municipalities and universities, as well as with various federations and associations for persons with disabilities.

The overall aim of Valteri is to provide equal opportunities for learning and development to all students. According to the role as a specialized center, Valteri supplements municipal and regional educational and habilitation services by offering a wide range of support services. The services may target the needs of individual students or the needs of an entire working community, municipality or region. The form, content and practical implementation of support services vary according to the individual needs. The most common services are counseling and consulting services for day-care centers and schools. These include periods with support for assessment, education and rehabilitation services for children and young people with special educational needs, the production of supplementary material for students with special needs, as well as production of publications and teaching material for professional staff. Valteri also organizes training courses or packages and seminars for professional staff working with students who have special needs.

The services provided for students with JNCL and their teachers and other professionals are offered nationwide from the Valteri centers. According to the Law on Financing Education and Culture, the services from Valteri are free for

guardians and partly paid by the education department of the municipality. The rest of the costs are covered by the State.

Valteri-Onerva has appointed a special education teacher who works nationwide as a counselor who is supporting the education of students with JNCL. This person has a close cooperation with central hospitals, the NCL professional group at the Finnish Federation for Visually Impaired and the Finnish NCL Family Association.

To start the pre-school path for a child with special needs and challenges, the family might contact the counselor, or the counselor may contact them. After discussion with the parents and having received their permission, the counselor contacts the rehabilitation worker in the regional hospital, the child's school teacher and the other professionals needed to get a comprehensive picture of the situation and to be able to plan how to proceed. The aim is to build up a supportive multi-disciplinary team for the needs of the child with JNCL.

Support for the pupil, teacher and multidisciplinary team at school level also takes place during the consulting visits. The consulting teacher observes the situation at kindergarten or school class and supports the teachers and others to proceed with the specific needs in education. At the end of the day, the parents, the multi-disciplinary staff and the rehabilitation workers usually have a joint meeting and share the observations of the consulting day. After each visit the consulting teacher writes a summary of the meeting, which is delivered to parents and the multi-disciplinary team around the child.

Valteri-Onerva also organizes in-service training for teachers and teaching assistants working with students with JNCL. The themes addressed and the extent and implementation of the training, as well as the costs, vary according to the type of training.

Special courses for students with JNCL are organized to support their education. These courses are mainly organized at Valteri-Onerva as group sessions for one school week. The students are gathered mainly according to their school grades once or twice a year. Each course includes individual assessment of special needs, such as functional vision, braille skills, computer skills, and mobility and orientation skills. The students are also given individual guidance and practice in adapted study techniques. The course package comprises teaching, learning materials, rehabilitation, leisure time activities, board and lodging. After the course, a multi-disciplinary report with possible recommendations for equipment and educational strategies, is sent to the parents, the school, and often to other parties involved.

According to the individual situation, it is possible to apply for school placement at Valteri-Onerva special school. The responsibility for organizing education rests primarily with the municipality, and negotiation between the local

educational authorities and the family is therefore needed before an application can be sent to the state special school. Valteri-Onerva also requires the child to attend the assessment course before accepting a possible school placement.

Whether services for a student with JNCL and his or her parents and teachers are organized locally or on the national level, it is crucial to focus on the multi-disciplinary work. The Finnish Federation for the Visually Impaired has an NCL working group. This team offers thorough professional support for families in cooperation with schools and rehabilitation units. The Finnish NCL Family Association is also an important part of this cooperation. As a peer support, they provide guidance and meetings for families as well as updated information on their internet pages.

Denmark: The Danish NCL Team

Denmark has a size of 43,090 square kilometers and a population of 5.8 million (2017). Copenhagen is the capital with 1.3 million inhabitants.

The multidisciplinary NCL Team in Denmark has six members: one social worker, two special education consultants, one doctor and two parents representing NCL Denmark (the Danish NCL Family Association). The multidisciplinary aspect is very important, from the time of the diagnosis. The close collaboration between medical, pedagogical and social counseling services, in conjunction with parents, continues throughout the life span. Parents of individuals with JNCL are valued for their expertise. The multi-disciplinary approach is important because the provision of services is based on observations and ideas informed by insights from many perspectives. The methods applied by the professionals are continuously discussed and elaborated with the team's parent representatives (Spielmeyer-Vogt Team, 2015).

Organization of team activities

In addition to its ordinary tasks, the NCL Team is concerned with organizational and professional issues. The relationship between professional knowledge and parental expertise is important, as well as the ways to maintain effective and sustainable networks on many levels: the children's network, the parents' network and the professionals' network. Counseling and supervision are provided locally in the municipalities where the children and their families live. The NCL Team emphasizes the importance of establishing a professional network around each child with JNCL, involving local services and parent participation.

In Denmark, the municipalities are responsible for the care of people with special needs, including those with rare diseases. The welfare system in Denmark allows parents to choose to keep their child with JNCL (or another form of NCL) either at home or in a public residence. Whatever the choice, the person with JNCL is guaranteed to receive an equal amount of public support. Young adults with JNCL receive a social pension beginning when they reach the age of 18 years. Most young adults with JNCL have their own apartment connected to the parents' home. The municipality provides pedagogical and nursing staff in their homes, as well as in schools and activity centers.

The NCL Team represents a supplement to the services offered by the municipalities and is organized and funded by the government. The team has nation-wide responsibility and is located at Synscenter Refsnæs, a regional center for visual impairment (see also Chapters 24 and 26).

The purpose of supervision

The NCL Team offers supervision and guidance to staff in the parental home. The Team also serves staff in schools, residential homes and activity centers. The competence of the local staff related to JNCL differs considerably. Some staff members have never received supervision and guidance while others are more experienced and have received supervision and attended courses.

The starting point for supervision is of particular importance. The nature of the disease can be overwhelming for staff with no experience. Early supervision will focus on possibilities, good teaching and quality of life, rather than on the disease. Early supervision is concerned with building a good culture and awareness of possibilities and appropriate initiatives, including the separation of professional and private feelings. The supervision will support the staff's communication with parents. Staff in parental homes or institutions will sooner or later meet the parent's dilemmas or feelings related to having a child with JNCL. Such situations will require professionalism by the staff and the NCL Team can support the staff in such situations. This is illustrated in the following story:

A staff group at a day care center communicated that they were worried about the contact between a mother and her son. During visits they observed the mother sitting passively beside the son doing nothing. There were no physical or communicative contact between the mother and her son. The staff questioned if the mother had any interests in her son.

The supervisor of the Team asked the staff members if they had discussed the situation openly with the mother and they answered no. The supervisor also asked if there could be other circumstances that should be considered

in relation to their observations. The staff members had never made such considerations.

In agreement with the staff, the supervisor from the Team talked with the mother about how she experienced the visits. The mother replied that she often became sad during these visits, because she did not have the same close and good contact as she used to have with her son. The supervisor asked if there were any particular causes for this change. The mother replied that she had been afraid of touching her son, because it might trigger epileptic seizures and anxiety.

The Team organized a meeting with the mother and the staff. It became evident that the mother had a need for guidance from the staff. The meeting resulted in a better and closer relationship between the son and the mother, and she became encouraged to obtain physical contact and communicate with her son as she had done in the past.

After the meeting, the staff became more concerned about how to interpret and meet parents' feelings. They would approach parents instead of making their own speculations. The following question became important: how would you like to be met in a similar situation?

The Team also participates in planning in the later phases of the disease. The purpose of the services is to relieve parents of the burden of practical care, so they can relate to their grief in peace, to the extent that is possible. This service is highly appreciated by the parents. To be a member of the Team entails more than just being a consultant to parents and staff. The relations between the Team and families become close over the years, and participation in difficult situations becomes a mandatory issue.

Meeting people's emotions

The Team's supervision is based on consideration and reflection. Supervision will focus on the process and understanding of actions, relationships, professionalism and individual responses when a child or young person is affected by losses. The supervision will also address the staff's and the parents' emotions that may affect the work with the person with JNCL. The supervisor's most important task is to act as a catalyst when emotions are affecting the daily work with the individual with JNCL. The Team meets the staff every third month on average, to share their experiences from the daily work. Evaluations show that these meetings are important to maintain the staff's spirit and engagement in the daily work.

Case story: Learning for Life

This story is written by a mother of a daughter with JNCL named Sofie. The usual length of education in Denmark is 13 years (primary and secondary school), but Sofie was not able to conclude her education in a traditional way.

Our daughter Sofie got problems when attending her 12th year of school because of seizures, bad days and lack of educational content. We, the parents, had to find an alternative educational solution for her. This short story is about this alternative school solution, tailored to meet the needs of our daughter Sofie.

After some discussion the municipality finally agreed to pay for home-based education for Sofie. This included full time staff (two persons) and miscellaneous costs, such as transportation. The home-based education saved money for the municipality and worries for us. Sofie was given full time support with two assistants from 8 am to 4 pm, Monday to Friday.



Sofie and her educational assistant.

We, the parents, in collaboration with the staff defined the main goals for Sofie's education:

- *Education and learning should be based on Sofie having a meaningful life with a strong focus on educational themes of importance for her.*
- *The educational activities should be based on selected themes.*
- *The school days should be structured, that is, follow a strict program to emphasize predictability.*

It was decided to have planning and evaluation meetings every second week to monitor education and create the best possible educational life for Sofie. Examples of selected themes were seasons, animals, favorite food, family and friends, love, the world, outdoor life, religions etc. These themes were converted to educational activities, for example:

- *The world theme was converted into specific activities, such as salsa dancing, African music, Thai food and stories about explorers.*
- *An animal theme was about lions. This theme was converted into activities like going to the Zoo and "seeing" a lion, roaring like a lion, watching and listening to the film The Lion King, smelling a lion, touching a toy lion, and making a lion steak.*
- *The outdoor life theme was converted into the following educational activities: Visiting the Queen's castle, beach tour, barbeque in the garden, visiting the rose garden, and taking a walk in the forest.*
- *The religion theme was converted into activities like learning and singing hymns, praying the Lord's Prayer, going to church, commune with crispbread and squash, baking, visiting Camilla's cemetery and eating fish and bread.*

One theme with many different activities could last for a week or more. The themes were addressed in a rather fixed schedule to promote best possible predictability as shown in Figure 9.1.

The home-based education worked very well for Sofie. She was engaged from early morning to late afternoon, and she learned a lot. The devoted staff became very familiar with Sofie and her special needs. Sofie's last years of education became a success for both Sofie and us, the parents.

Date	12th September Monday	13th September Tuesday	14th September Wednesday	15th September Thursday	16th September Friday
Theme	Theme Africa				
08.00–09.00	Breakfast, plan for the day and song				
09.00–10.00	African music and dances				
10.00–11.00					
11.00–12.00	Prepare African lunch				
12.00–12:45	African lunch				
12.45–13.00	Cleaning the kitchen				
13.00–14.00	Tour to Zoo to watch and listen to African animals	Tour to Zoo to watch and listen to African animals	Tour to Zoo to watch and listen to African animals	Tour to Zoo to watch and listen to African animals	Tour to Zoo to watch and listen to African animals
14.00–15.00					
15.00–16.00	Summary of today's activities and massage				
Compulsory activities					

Figure 9.1 Sofie's school schedule for one week.

Norway: The structure and function of services

Norway has a size of 385,180 square kilometers and a population of 5.3 million (2017). Oslo is the capital with 674,000 inhabitants.

The public services in Norway are based on a law that says that all citizens have the same right to services, independent of who they are and where they are living (Hesselberg, 2011). However, equality in services is difficult to achieve in a country like Norway because the population is scattered and the number of inhabitants in the municipalities varies from 500 to 674,000 inhabitants.

There are three administrative levels in Norway: The municipalities are responsible for kindergartens, primary and secondary schools and special education, including special education for adults. Health services on the local level include health centers, physiotherapists, general practitioners, community care institutions and school psychology agencies. The counties are responsible for transport,

dentists, secondary education with school psychology services, and education in different institutions. The national level, the State, is responsible for specialized health services, centers for rare diseases, special training centers, centers for special education, labor and welfare administration, technical aids and special interest organizations. Persons with JNCL are receiving services from all these levels.

Most children with JNCL in Norway attend mainstream schools. There are very few special schools in Norway and no special school for students with visual impairment. Some mainstream schools have special units for students with special needs, which may include for example children with intellectual disabilities, severe motor impairment or autism spectrum disorders. Adults with special needs may be provided with special education after assessment, recommendations and case considerations by the local or regional educational authorities. This right to education is not limited in time or age.

The developmental course of JNCL creates challenges for the health and education department of the municipalities when providing education and other interventions. The local professionals will meet a person with a rare progressive disease they have never heard about. Life-long changing needs will require commitment and collaboration between the parents, the schools and other public institutions. This collaboration is particularly challenging because it cannot be based on a standard system but must be based on a person-centered approach (see Chapter 11).

The services have to adapt to the needs of persons with JNCL, which are increasing with age, and there are considerable differences between persons in this group. Different institutions are responsible for providing the services and competence building needed to support persons with JNCL. The local and specialized services must be coordinated, and there is usually a formal agreement on responsibilities and cooperation. There is a need for support and guidance for those who are close to the person with JNCL, but also for other people in the environment.

The workshop model

All services must be based on sufficient and relevant knowledge. Knowledge about JNCL is limited in general and is not usually found among professionals working in the municipality or in county-level institutions in Norway. Building up local knowledge and competence is a core issue in provision of educational and other services for individuals with JNCL.

A person-centered approach requires two types of insights: knowledge about the person with JNCL and knowledge about JNCL. Knowledge about JNCL is mainly found on a national level, in specialized centers or resource centers. Some of these institutions are providing outreach services, such as Statped.

Knowledge about the person is well represented locally, in the family, the school, and sometimes within local or regional educational counseling services. Statped Midt in Trondheim has specialized competence related to education of students with JNCL and is usually responsible for building up the necessary local competence when a new child is diagnosed with JNCL. Statped midt cooperates with the family, the local school and pedagogical-psychological services, and other local or regional services. It is necessary to establish a close and secure relationship and efficient ways of communication, tailored to the needs of the child or young person with JNCL. Figure 9.2 describes a workshop model for collaboration that has proved useful in many cases.

There is always a risk that the challenges related to JNCL can seem overwhelming for staff receiving competence-building services. The workshop model allows the inclusion of bottom-up strategies where competence building and guidance to some extent is based on everyday situations, situations that are successful as well as with negative experiences. The workshop model and the bottom-up strategy invite the participants to set the agenda of the workshop and address topics that are important to them.

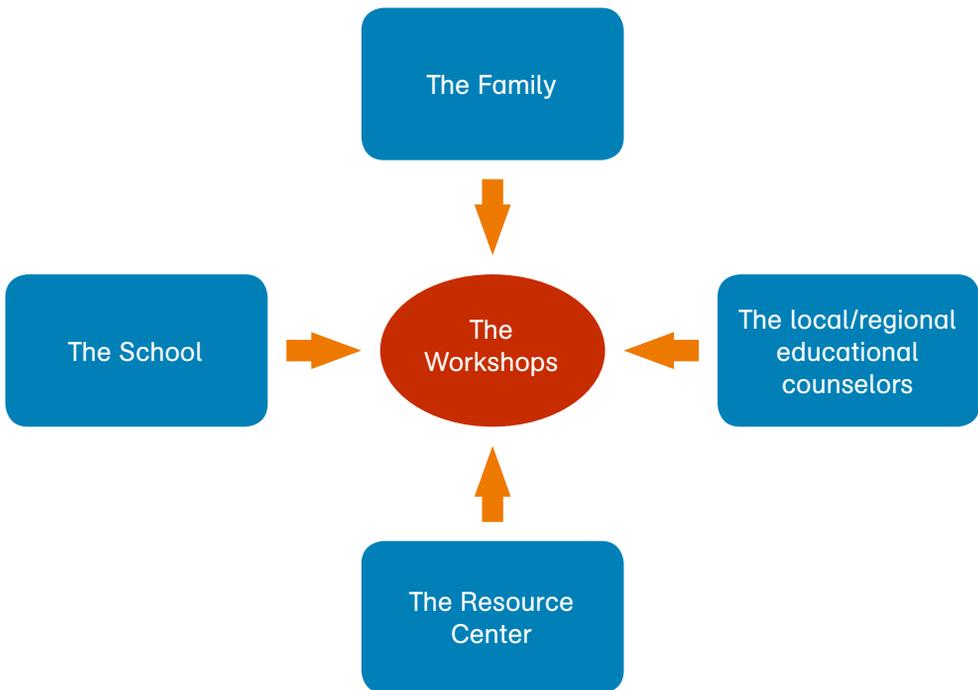


Figure 9.2 The workshop model for person-centered collaboration

Experiences with applying this model indicate that the specialized professionals need to spend some time in the school and the home prior to the workshop. This provides an additional perspective as the JNCL specialist may observe and understand things that people with less experience with the disease are not likely to see. Further, the specialist observing the child or young person with JNCL in familiar surroundings and in different situations may get a more accurate understanding of the issues that are going to be discussed in the workshop.

The workshop usually takes one working day. If a longer workshop is planned, it may be difficult for the family to find time and for organizers to get all the relevant professionals together. Leaders and administrators may not be able to attend the whole workshop but are often able to come for the summing up and allocation of responsibilities. A written report with conclusions and recommendations is always provided by the JNCL counselor after the workshop.

In addition to workshops, Statped Midt organizes 3- to 4-day center-based courses for teachers, other staff and parents. An advantage of attending center-based courses is the possibility of meeting specialists who would not come to local workshops, and of meeting other professionals who are working with other individuals with JNCL. However, the attendance of center-based courses is often limited, possibly due to the expenses they represent for the municipalities.

The one-day local workshops seem more manageable for schools and other institutions. They are often attended by ten or more participants: teachers, residential staff, assistants and parents. A workshop thus reaches a substantial group of the people around the person with JNCL. Workshops are usually managed by one or two professionals who have specialized on JNCL. An important argument for local competence building is the fact that one can concentrate on the issues related to just one individual, which is the foundation of the person-centered approach. The courses have a more general content and may not meet the knowledge needs of all the parents and professionals who attend the course.

The Norwegian experiences with applying the workshop model are good. However, the model requires competent counselors who can meet the local needs, which may vary considerably with the person's age, development and declines, as well as interests and personality. It takes time to build such competence and it requires access to the diverse and various fields of specialization that are involved in the education and habilitation of this group. There is at present no formal education specializing in the educational needs of persons with JNCL. The Norwegian national educational competence on JNCL is built through in-service training.

References

- Education (Additional Support for Learning) (Scotland) Act 2004. Retrieved 2017 from <https://www.legislation.gov.uk/asp/2004/4/contents>
- Hesselberg, F. (2011). Fleksibel utnyttning av lokal fagkompetanse (Flexible utilization of local professional competence). In S. Strand (Ed.), *Samhandling som omsorg*. Oslo, Norway: Kommuneforlaget.
- Scottish Executive (2002). Standards in Scotland's Schools Act 2000. Guidance on Presumption of Mainstream Education. Retrieved 2017 from <http://www.gov.scot/Resource/Doc/46922/0024040.pdf>
- Scottish Government (2010) A guide to implementing *Getting it Right for Every Child: Messages from pathfinders and learning partners*. Edinburgh: Scottish Government. Retrieved October 2018 from <https://www.gov.scot/Resource/Doc/318937/0101828.pdf>
- Snunit, M, & Golomb, N. (1991). *Der Seelenvogel*. Hamburg, Germany: Carlsen.
- Snunit, M, & Golomb, N. (1999). *The soul bird*. New York, NY: Hyperion Press.
- Spielmeyer-Vogt Team (2015). *Pedagogiske overvejelser i arbeidet med børn og unge med sygdommen Spielmeyer-Vogt* (Pedagogical considerations in the work with children and young people with Spielmeyer-Vogt disease). Kalundborg, Denmark: Synscenter Refsnæs.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.

10

Assessment and Evaluation

*Stephen von Tetzchner, Anne-Grethe Tøssebro, Heather Adams,
Barbara Cole and Bengt Elmerskog*

Evaluation is an ordinary part of all education, in the form of grades or qualitative feedback. The assumption is that feedback helps students understand what they need to learn and guides them toward learning strategies that may support their learning (Baird, Andrich, Hopfenbeck, & Stobart, 2017; William, 2018). Assessment goes beyond an evaluation of the student's actual school work and is to a larger extent a help to those who are responsible for the education. An assessment gathers information about how the student understands, thinks, feels and acts in relevant areas, and on the characteristics of the physical and social environment, including relations within the family and with peers and adults outside the family. The information should indicate whether an individual has difficulties in areas requiring treatment, special education or other measures, or if changes need to be made in the individual's environment. The function of assessment is thus to establish a foundation for making decisions about intervention, including the decision that intervention is not necessary.

Assessment is a process that is usually initiated on the basis of a concern and a referral, and these will guide the assessment. The referral identifies the concern; the function of the assessment is to find out *what* the problems might be and *why* the child is showing these problems. When the diagnosis of the child or young person is known, knowledge about the typical course and variation in symptoms within the diagnosis will guide what domains to assess. In addition, assessment will build on the strengths and weaknesses observed in earlier assessments. The developmental and learning histories of the individual give information about the problems encountered by the individual and how the interventions functioned, including, for example, to what extent they led to increased knowledge and independence, or enabled a student with declines to continue doing things with help. They also inform about growth and decline in various domains, which may be helpful in adapting new intervention measures to the individual's present functioning and assumed functioning in a near and more distant future.

Although children and young people with JNCL share many of the same difficulties, there are also significant differences between their developmental patterns. For example, not all individuals show a full range of known JNCL problems, and the symptoms are not manifested at particular ages or in set order. This diverse profile is evident in case presentations discussed in many chapters of this book. Thus, to regard individuals with JNCL as a homogeneous group would mask important differences in their possibilities, limitations, their coping mechanisms, and their respective need for support and environmental adaptations. Assessment is therefore essential for capturing these important differences so they can be used to customize shorter and longer term treatments and life plans even if the overall long term course of the disease is the same.

A review of assessment in general is beyond the present chapter (see for example Brassard & Boehm, 2007; Crick, 2007; Dombrowski, 2015; Sattler, 2008a,b; Williams & Hill, 2012). Instead, this chapter describes only the basic assessment strategies and processes relevant to the assessment of individuals with JNCL. It also presents a checklist for observing developmental progress and decline in these individuals the *Educational Development Observation Tool (EDO)*, which has been designed as part of the JNCL and Education Project.

Assessment strategies and processes and JNCL

Assessment may include direct and indirect measures. Direct measures come from tests and observations while indirect measures are based on information from structured conversations or standardized interviews with parents, teachers or others about the student's functioning in specific areas. The conversations provide important information about the student and the family but also contribute to making professionals, teachers and parents acquainted with one another and to establish an *alliance*, that is, a shared understanding of the student's strengths and weaknesses and the types of measures that may be useful. The conversations also provide information about the student's physical and social surroundings, about the parents' and the teachers' opinions about earlier and present intervention measures, and about resources and deficits in the student's environment. Each profession has its instruments and strategies for assessment. Feedback to parents and teachers from assessments and observations is also typically mediated in a conversation (Hesselberg & von Tetzchner, 2016).

In standardized interviews, there is a tendency for the professional to ask all the questions, and indeed to set the agenda through their prechosen question set, and for the parents' role to be relegated to answering those predefined questions. Structured and semi-structured conversations are more open, thus allowing and

encouraging both parents and professionals to introduce topics freely, according to how the topics arise in the conversation. A guiding principle for conducting both conversations and interviews is the importance of creating an open and secure situation for all participants. This principle is particularly important when the parents know more than the professionals, which is often the case with rare disorders like JNCL (see Chapters 24 and 26).

An observation procedure provides a goal-oriented perception of what one or several individuals are doing. Observations conducted in the home or at school can give the professional an impression of the student's functioning in ordinary situations, including levels of attention, performance, effort, social functioning and participation. It can be useful to observe the student in both structured and unstructured situations, and together with both peers and adults. There are some checklists with predesigned categories for recording children's play and activities (Barton, 2010; Kelly-Vance, Ryalls, & Gill-Glover, 2002), but these may not be valid when the child has a disability (e.g., blindness or motor impairment) that interferes with performance in ordinary activities.

If testing is part of the assessment, then the tests must be appropriate for the level of the individual, and should be evidence-based (Hunsley & Mash, 2007; Bruce, Luckner, & Ferrell, 2018). Moreover, the validity of the instruments used in the assessment of a child or adult with a diagnosed or suspected disorder must be documented for that particular diagnostic group. In fact, the use of assessment instruments that are not appropriate for the person who is assessed is considered a major breach of professional ethics in school psychology (Leach & Oakland, 2007; Mendes, Nascimento, Abreu-Lima, & Almeida, 2016).

Although tests are designed to be suitable for most individuals, it may be necessary to adapt the test situation and the material being used to the perceptual and physical abilities of the individual (Zebehazy, Zimmerman, & Zigmond, 2012). Assessments of individuals with severe visual impairments are often based only on the verbal part of the test. Tasks consisting of images with a missing item, categorization based on shape and colors, puzzles and checkerboard patterns are unsuitable for children with poor vision. Many tasks used to measure language comprehension require ability to follow instructions that involve locating items on a table or in the immediate surroundings. These tasks also represent challenges for individuals who are blind. Some individuals with JNCL have motor problems, which will influence their ability to follow instructions that require motor performance. Moreover, problem-solving will vary with the familiarity of the material (Wason, 1977). A test with material that is very unfamiliar to the individual may be less useful to measure the individual's skills than tasks with more familiar material (see also Chapter 12). The problems related to test assessment of children with visual impairment are expressed strongly by Erin

and Koenig (1997): «The use of standardized tests with students who have visual disabilities is fraught with complex problems. Even with stringent and thorough adaptation, such tests alone rarely provide sufficient information for making important decisions about students with visual disabilities» (p. 311). However, there are also tests with non-visual non-verbal tasks for assessment of children with visual impairments (Smith & Amato, 2012; Zebehazy et al., 2012).

Any special arrangements and adjustments that are made during testing must be taken into account when interpreting the results. When testing takes place under specially tailored conditions, the norms may merely provide a guideline, since they are usually based on a representative group of children with typical development. Children with atypical development are rarely included in norm-referenced tests. Consequently, the norms may not apply and should be used with caution; nonetheless, they may still be useful for optimizing the children's learning situation.

Norm-referenced tests are used for ranking individuals and comparing them to their peer group. When it has been established that performance is much lower than expected for the age, the norms serve little purpose. The test scores in themselves are less important than the implications they may have for educational strategies (Lichtenberger, Mathen, Kaufman, & Kaufman, 2009). Criterion-referenced tests are used to assess what an individual can and cannot manage, with the aim of establishing what kind of training he or she needs to acquire the missing skills. Criterion-referenced test results thus indicate how closely the child is to skill mastery in some domain, instead of how the child compares to the peers. For students with JNCL, criterion-referenced tests may be useful in assessing degree of decline and amount of help needed.

Traditional use of tests is often referred to as "static", because the tests measure knowledge and skills at a particular point in time. By contrast, from a social constructivist perspective on cognition, assessment should record not only which tasks the child does and does not master (i.e., static appraisal), but also the learning process itself. Thus, from this perspective, assessment must be dynamic; dynamic assessment implies identifying the child's zone of proximal development – what the child is able to do with help, and how much help is needed to solve a given task (Jeltova, Birney, Fredine, Jarvin, Sternberg, & Grigorenko, 2007; Lidz, 1997). Dynamic assessment does not merely measure the individual's skills at a given time but what the individual can do with help and learn under standard conditions; thus, norms must be used with caution. Static assessment may be a way to assess independent function, and dynamic assessment a way to assess interdependent function and the kind of and amount of help that are needed, thus assessing the zone of developmental maintenance (see Chapters 2 and 16).

Cognitive assessment should never rely on tests alone. According to Erin and Koenig (1997), «The results from an assessment, whether standardized or

not, must never be the sole source of information on which to base any important educational decision» (p. 313). Test results should be collated with observations of how the individual copes with familiar and unfamiliar everyday situations. The teacher's observation may be useful but there are some limitations. For example, due to the low incidence of severe visual impairment in school-age children, teachers and other professionals in mainstream schools often feel inadequate in evaluating students with visual impairments (Loftin, 1997). Observations of the student in the classroom and other familiar surroundings may be useful in seeking to assess possible cognitive decline or dementia. However, observations are time-consuming and a short observation may not provide a full impression of the variation in the individual's functioning. A checklist may function quite well and take less time.

Checklists comprise questions about skills, behavior and everyday functioning, and have become a common element in assessment. They are usually completed by parents, teachers and other adults, but checklists can also be self-completed by older children and adolescents. A checklist can also provide a good basis for conversations about the child's functioning. In particular, in relation to behavior, there are often significant differences between the answers provided respectively by the parents, teachers, and the child or adolescent (De Los Reyes & Kazdin, 2005; Rescorla et al., 2014). However, it is important that parents should not be responsible for evaluating their child's development alone – they see the child through their special "parent glasses" and even the glasses of the mother and the father can be quite different (Seifer, 2002). Checklists can be part of the parent interview and contribute to a dialogue about the abilities of the child and the features of the environment (Rosenbaum & Valsiner, 2011).

In addition, checklists are used by professionals to structure their own observations. An advantage with checklists is that they give information about the everyday functioning of the individual, but like tests, it is important that they concern actions, skills and activities within domains that are relevant and categories that are appropriate for the person. Checklists may be a useful approach in assessing individuals with JNCL provided they are suitable for individuals who are showing visual and cognitive decline. The Educational Development Observation Tool (EDO) is especially designed for this group (see below).

Giving tests and completing checklists represent only the first stage in the assessment process. The main task is to analyze the results, describe functional profiles with strengths and weaknesses, and compare test results, observations and information from interviews and conversations. For example, the declines in language and cognition do not always start at the same point in time. Some children with JNCL develop speech problems (i.e., hypokinetic dysarthria) early while their cognitive functions, including linguistic skills underlying expressive

and receptive language remain seemingly unchanged for some time. For other children with JNCL, it is the other way around, they have problems with language while their speech is clearly intelligible. This implies that non-verbal cognition, expressive and receptive language, and motor speech functioning must be evaluated separately. When cognition is compromised, there will be difficulties with receptive and expressive language (i.e., the symbolic code, vocabulary, ability to understand ideas in complete sentences, etc.), but speech itself need not be affected. Alternatively, problems with motor speech production can mask an adequate language system. Problems with articulating speech should therefore not be interpreted as an indication of cognitive problems or decreased language comprehension. An individual with hypokinetic dysarthria might also simplify how ideas are put into words, in the interests of being intelligible to others. This simplification process can make it appear that the speaker lacks age-appropriate vocabulary or grammar, when in reality, the simplification is strategic, thus reflecting adequate cognition and judgment.

It is important to compare the individual's strengths and weaknesses as they emerge in a structured test situation with the individual's functioning in structured and unstructured activities at home, at school and in less familiar places. Dynamic assessment strategies may shed light on the processes of decline, thereby supplementing other observations used to assess the amount of help the individual needs to cope in various situations.

Educational initiatives for individuals with JNCL must build on an alternative principle compared to more traditional initiatives within special needs education, because decline, not progress, is expected over time. Standard evaluations focus on

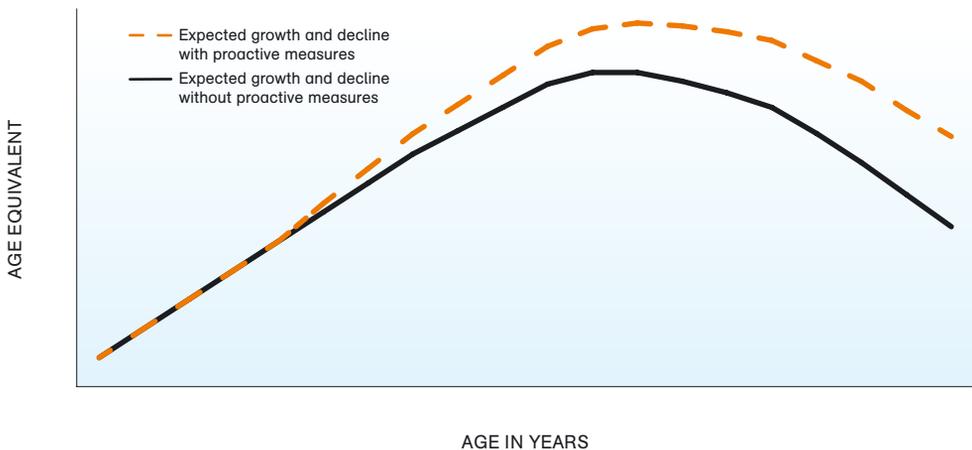


Figure 10.1 Hypothetical development with and without proactive intervention

successive improvements in personal performance and growth. This focus is also important for individuals with JNCL, but evaluation must be based on the extended perspective. Having JNCL will always imply declines and losses affecting the individual's level of performance and functioning. Results from the present JNCL project (Appendix A) indicate that appropriate educational initiatives may delay the onset of the descending curve on functions and slow down the rate of decline, thus leading to longer maintenance of functions and skills. The developmental trajectories in Figure 10.1 illustrate this extended perspective. The figure shows a hypothetical difference between a person with JNCL's growth and decline with and without proactive or preventive measures. The lower line illustrates growth and decline with no proactive measures while the upper line shows growth and maintenance with proactive measures.

The Educational Development Observation Tool (EDO)

The Educational Development Observation Tool (EDO) is designed to give an overview of eleven central themes or domains that should always be considered in planning for persons with JNCL. The EDO is a tool that can be used for long- and short-term assessments and interventions, and as a background to facilitate the development of the Individual Education Plan (IEP) and the Habilitation Plan (see Chapter 11). The central themes identified in the EDO are vision, communication, literacy, social life, gross motor function, fine motor function, physical activity, behavior, memory and attention, independence and autonomy, interests and equipment. These themes can be met by different interventions. Some themes included in the EDO, such as expressive communication are addressed from a long-term perspective, because the use of speech and written communication will decline sooner or later; other domains must be considered in the short term, and the EDO helps identify what can be done now to meet known future challenges.

The EDO tool is tailored for individuals with JNCL and includes assessment domains and suggestions for interventions based on the assessment findings. The tool focuses on individual functions and environmental factors. Parts of the tool can be used for other groups with disabilities, for instance for children with other diseases involving dementia. The main aim of the EDO is not mapping declines or symptoms but identifying the best possible interventions to optimize quality of life despite all challenges. The EDO's three broad objectives are (a) to support educators, social workers, residential staff, parents and others in achieving the best possible quality of life for individuals with JNCL, (b) to identify interventions to

meet current or future challenges, and (c) to create a focus on possibilities instead of barriers. The EDO should be used to support development, learning, maintenance and teaching in and beyond the classroom, and within and beyond school age.

The EDO has been developed as a part of the *JNCL and Education Project* (see Appendix A). The body of experiences obtained from the use of the EDO was limited when this book was published, and it is assumed that the EDO will undergo further development and refinement in the coming years.

Use of the EDO

The EDO is an interactive document. It is extensive in size, but rather time-efficient to administer when one knows how to use it. It is recommended that it be used annually as a preparation for the next operational year, such as the next school year. It is particularly important to use the EDO prior to major transitions, such as transitions between schools, or the transition into adult living.

The tool should be completed by the parents together with a team composed of educationalists, social workers, JNCL specialists and possibly others who know the person well. The time constraints under which parents and professionals operate are important considerations. Thus, the JNCL specialist should have sufficient knowledge about the EDO and be proficient in its administration, including being able to complete the tool efficiently, ensuring that the assessment does not take more time than necessary.

The assessment part should be based on the person's current optimal functioning, that is, when the person with JNCL has a "good day" in an optimal context. However, it is also important to know how the person functions during periods when he or she is affected by seizures or a negative mood, or is in unfamiliar or nonpreferred settings.

Some questions or topics of the EDO may not be considered relevant at all times, depending on the person's current situation. The decision to leave out topics or questions should be made in close collaboration with the JNCL specialist. Another important role of the JNCL specialist is to ensure that the team consider early educational interventions that are not currently obvious, but that might be beneficial in the future. The JNCL specialist should provide guidance about expected future symptoms, and how to prepare the person with the JNCL disease to meet these symptoms in best possible way.

JNCL includes a series of declines in cognition, mobility, vision, communication and independence. Educational interventions cannot stop the declines, but education can optimize interventions and compensate for some losses and problems caused by the declines. Children, adolescents and young adults with JNCL should – for as long as this is possible – follow the ordinary curricula and

participate in everyday, socially appropriate activities even if these are not clearly illustrated in the EDO. The EDO focuses on educational areas that are specific to individuals with JNCL and that will require interventions that are not applied in the education of the typically developing population. The EDO is based on educational strategies and principles described earlier in this chapter and other chapters of the book, such as hastened learning, proactive learning, skill-oriented learning, participation-based learning and life flow.

A brief introduction of the content

The EDO has 12 parts or domains of particular importance for individuals with JNCL. All parts are important, but all questions may not be important at all times. The questions or topics in each part should give a picture of the person's present situation (the observations) followed by an analysis of the consequences for education and non-medical service provision (the interventions). The questions may sometimes be difficult to answer but should still be elaborated as well as is possible. An overall objective when using the EDO is to ensure that persons with JNCL will receive appropriate services and interventions at the right time.

Part One: Demographic information. The team collects personal details such as name, age, where the person lives, school and who participated in the completion of the EDO.

Part Two: Vision. The vision of children with JNCL is normally assessed by ophthalmologists. This part gives a brief description of the visual function that may be of importance for non-medical interventions. The change from being a person with functional vision to become a person who is blind is a difficult transition. Such concerns as losses (e.g., friends, favorite activities) and the growing need to complete tasks in new ways must be considered by educational planners. It may also be desirable to prepare the child for the forthcoming visual impairment by using pro-active learning in certain areas. The onset of visual impairment is typically evident when the children receive the JNCL diagnosis. Visual impairment may imply the selection of some new goals and new adaptations. With the onset of blindness, more goals may need to be revised, and optimization and compensation are likely to be comprehensive and wide-ranging, and hence requiring insight into both blindness and the other features of JNCL. Preparation for blindness implies making the student's surroundings blind-friendly, by removing hazards, marking important places, and providing the environment with tactile landmarks, shorelines and so forth (see Chapters 4 and 16).

Part Three: Communication. The JNCL disease will – eventually – include severe problems with speech. The person's language comprehension will often remain better than the speech intelligibility. Education should consider how to

support current or forthcoming speech problems by using enhanced and proactive learning. Both speech and language therapy and the use of alternative and augmentative communication forms (AAC) should be discussed with a JNCL specialist in an early phase of the disease, even if the student's speech at this time is functioning well in all situations (see Chapters 6 and 13). However, there is a need to collect more evidence and build competence from systematic training of AAC for students with JNCL. This is an important area where future research is needed.

Part Four: Literacy. Reading and writing are pivotal skills. Most children with JNCL will at first learn reading and writing through the visual modality. Many of them are eventually offered learning of tactile reading and writing. Delaying the introduction of tactile reading and writing may make learning difficult due to the concurrent, progressive cognitive decline. It is advisable to consider tactile reading and writing when the child's learning capacity is at its best, even if the child's sight still is sufficient for visual reading and writing. It is recommended that decisions about reading and writing instruction are made in close collaboration with JNCL specialists (see Chapter 14).

Part Five: Social life. Social life is of great importance for everyone, but it can be affected negatively by the many symptoms of JNCL (e.g., declines in vision, communication and motor skills; and possible changes in mood and behavior). Problems will, to some extent, reflect shortcomings in the environment and may be reduced when environmental adaptations are made to meet the person's social and learning needs. The ability to participate in social settings can be facilitated by inclusive interventions, by using the interactive team model, by providing support and other environmental measures (see Chapter 22).

Part Six: Gross motor function. All persons with JNCL will be affected by gross motor declines. The motor declines may restrict participation in certain life situations if not met by appropriate interventions. Interventions (e.g., physical adaptations in the environment, provision of supporting materials, technical aids etc.) will reduce the effects of the declines. Anticipating forthcoming motor declines is the best way to ensure that each person with JNCL can achieve a good outcome (see Chapters 7 and 15).

Part Seven: Fine motor function. Because persons with JNCL become blind, they come to rely upon use of their fingers to explore the world. Fine motor coordination is a necessary skill for performing and participating in every-day activities such as eating, cooking, reading, and so forth. In the early phases of disease, while children are still capable of learning and mastering new skills, fine motor skills should be trained and developed. As the disease progresses, fine motor functions should be maintained to highest possible extent, and limitations should be addressed through the provision of customized task adaptations.

Part Eight: Physical activity. Motor problems and visual impairment will affect options for being physically active. Physical fitness is therefore an important goal (see Chapters 7 and 15). Being physically active is of special importance for individuals with the JNCL disease because through such activity, overall functioning can be preserved and declines mitigated. It is desirable that persons with JNCL are physically active every day. Physical activity includes participating in sports and dancing, and it can be incorporated into daily routines such as walking to school. Physical activity should be supplemented with physiotherapy when needed. It is important to be in good shape to meet future motor declines. The IEP and Habilitation Plan should include a training program and regular activities that involve physical performance that is adapted to the motor functioning of the person.

Part Nine: Behavior, anxiety and mood. Behavior problems, worries, anxiety, problems with mood and so forth are known challenges for some, but by no means all individuals with JNCL (see Chapter 27). These problems often occur after the onset of severe visual impairment and in combination with dementia. Challenging behaviors, worries and anxiety can be difficult to handle and may in turn affect learning and interaction with peers. Such conditions may subside if appropriate measures are taken. The IEP and Habilitation plans should include plans for environmental adaptations as well as appropriate reactions from family and staff.

Part Ten: Attention and memory. Attention and memory are two vital components for learning and performance. All individuals with JNCL will sooner or later have problems with their attention span and the ability to remember new things. Long-term memory is considered to be an important resource for persons with JNCL, whereas working memory is more affected by the disease. Thus, there is a need to address such problems with educational strategies that are appropriate at each phase of the disease (see Chapter 12); moreover, it is important that teachers and educational staff are alerted to potential for memory problems and receive the training needed for implementing useful strategies as the need arises. Examples of interventions include: providing enough repetition, structuring, and clear information; asking closed-set questions instead of open-ended questions; and implementing memory lists.

Part Eleven: Independence and autonomy. In their early years, persons with JNCL are just as independent as their peers. The declines occurring later will decrease their ability to act independently. Developing and maintaining independence should always be considered within education planning whenever possible. Autonomy must be reconceptualized from the perspective of what it means to maintain some sort of autonomy in the face of the many and inevitable functional declines. The interactive team model (see Chapter 16) provides a suitable framework within which a team should successively compensate for the inevitable

decrease of independence caused by different declines. This model, illustrated in the EDO, focuses on what the person can participate in when supported by a helper. Early planning and preparedness are important for maintaining independence by teaching new behaviors and making adaptations in the environment (see Chapter 16). The planned interventions should facilitate an understanding of the environment that is based on earlier knowledge and available sensory information.

Part Twelve: Interests and equipment. This part can be used to identify activities important for the person's quality of life and ensuring best possible life flow. The part can also be used to specify equipment that has a positive impact on the person's life situation.

References

- Baird, J. A., Andrich, D., Hopfenbeck, T. N., & Stobart, G. (2017). Assessment and learning: Fields apart? *Assessment in Education: Principles, Policy and Practice*, 24, 317–350.
- Barton, E. E. (2010). Development of a taxonomy of pretend play for children with disabilities. *Infants and Young Children*, 23, 247–261.
- Brassard, M. L., & Boehm, A. E. (2007). *Preschool assessment: Principles and practices*. London: Guilford Press.
- Bruce, S. M., Luckner, J. L., & Ferrell, K. A. (2018). Assessment of students with sensory disabilities: Evidence-based practices. *Assessment for Effective Intervention*, 43, 79–89.
- Crick, R. D. (2007). Learning how to learn: The dynamic assessment of learning power. *Curriculum Journal*, 18, 135–153.
- De Los Reyes, A., & Kazdin, A. E. (2005). Informant discrepancies in the assessment of childhood psychopathology: A critical review, theoretical framework, and recommendations for further study. *Psychological Bulletin*, 131, 483–509.
- Dombrowski, S. C. (2015). *Psychoeducational assessment and report writing*. New York: Springer.
- Erin, J. N., & Koenig, A. J. (1997). The student with a visual disability and a learning disability. *Journal of Learning Disabilities*, 30, 309–320.
- Hesselberg, F., & von Tetzchner, S. (2016). *Pedagogisk-psykologisk arbeid* (Pedagogical-psychological work). Oslo, Norway: Gyldendal Akademisk.
- Hunsley, J., & Mash, E. J. (2007). Evidence-based assessment. *Annual Review of Clinical Psychology*, 3, 29–51.
- Jeltova, I., Birney, D., Fredine, N., Jarvin, L., Sternberg, R. J., & Grigorenko, E. L. (2007). Dynamic assessment as a process-oriented assessment in educational settings. *Advances in Speech Language Pathology*, 9, 273–285.
- Kelly-Vance, L., Ryalls, B. O., & Gill-Glover, K. (2002). The use of play assessment to evaluate the cognitive skills of two- and three-year-old children. *School Psychology International*, 23, 169–185.
- Leach, M. M., & Oakland, T. (2007). Ethics standards impacting test development and use: A review of 31 ethics codes impacting practices in 35 countries. *International Journal of Testing*, 7, 71–88.

- Lichtenberger, E. O., Mather, N., Kaufman, N. L., & Kaufman, A. S. (2009). *Essentials of assessment report writing*. London, UK: Wiley.
- Lidz, C. S. (1997). Dynamic assessment approaches. In D. P. Flanagan, J. L. Genshaft & P. L. Harrison (Eds.), *Contemporary intellectual assessment. Theories, tests, and issues* (pp. 281–296). London, UK: Guilford Press.
- Loftin, M. (1997). Critical factors in assessment of students with visual impairment. *RE: View*, 28, 149–159.
- Mendes, S. A., Nascimento, I. M., Abreu-Lima, I. M., & Almeida, L. S. (2016). A study of the ethical dilemmas experienced by school psychologists in Portugal. *Ethics and Behavior*, 26, 395–414.
- Rescorla, L. A., Boichichio, L., Achenbach, T. M., Ivanova, M. Y., Almqvist, F., Begovac, I., Bilenberg, N., Bird, H., Dobrean, A., Erol, N., et al. (2014). Parent–teacher agreement on children’s problems in 21 societies. *Journal of Clinical Child and Adolescent Psychology*, 43, 627–642.
- Rosenbaum, P. J., & Valsiner, J. (2011). The un-making of a method: From rating scales to the study of psychological processes. *Theory and Psychology*, 21, 47–65.
- Sattler, J. M. (2008a). *Assessment of children: Cognitive foundations, Fifth edition*. La Mesa, CA: Sattler Publisher.
- Sattler, J. M. (2008b). *Resource guide to accompany assessment of children: Cognitive foundations, Fifth edition*. La Mesa, CA: Sattler Publisher.
- Seifer, R. (2002). What do we learn from parent reports of their children's behavior? Commentary on Vaughn et al.'s critique of early temperament assessments. *Infant Behavior and Development*, 25, 117–120.
- Smith, D. W., & Amato, S. (2012). Synthesis of available accommodations for students with visual impairments on standardized assessments. *Journal of Visual Impairment and Blindness*, 106, 299–304.
- Wason, P. C. (1977). The theory of formal operations: A critique. In B. Geber (Ed.), *Piaget and knowing: Studies in genetic epistemology* (pp. 119–135). London: Routledge and Kegan Paul.
- William, D. (2018). How can assessment support learning? A response to Wilson and Shepard, Penuel, and Pellegrino. *Educational Measurement: Issues and Practice*, 3, 42–44.
- Williams, J., & Hill, P. (2012). *A handbook for the assessment of children's behaviors*. Oxford, UK: Wiley.
- Zebehazy, K. T., Zimmerman, G. J., & Zigmond, N. (2012). Considerations in adapting alternate assessments for students with visual impairments. *Insight: Research and Practice in Visual Impairment and Blindness*, 5, 123–131.

Planning Education and Intervention for Individuals with Juvenile Neuronal Ceroid Lipofuscinosis (JNCL)

*Bengt Elmerskog, Stephen von Tetzchner,
Anne-Grethe Tøssebro and Finn Hesselberg*

An essential element for developing and sustaining modern societies is the existence of a system of education for all. Schools are important societal institutions for giving children and young people the knowledge they need, and in accordance with the needs of society (Ballantine, Hammack, & Stuber, 2017; Hesselberg & von Tetzchner, 2016). All modern societies therefore have both an organizational structure for education and general plans for all levels of education (i.e., primary, secondary, colleges and universities). During early school years, most individuals attend the local primary school where the curriculum has little scope for personal choice, and any choices tend to be made by the parents. In higher school grades and university, there are more opportunities for individual choice among the courses offered, and because the children are getting older, they are given more latitude in making their own choices.

While a large majority of children and adolescents follows the ordinary or mainstream curriculum, some students and young people have disorders and disabilities that necessitate a more individualized educational course. However, like other students they also build knowledge and skills, although usually at a slower pace or in atypical ways.

The educational situation is very different for students with JNCL. While their knowledge and skills in the first years increase more or less similarly to their peers, the perceptual, motor and cognitive declines will gradually reduce their ability to search for, process and convey information. Their understanding of the physical and social world, their own needs and their current and future possibilities will gradually decrease (see Chapters 4–7). They will need more help in communication and everyday functioning. They become dependent on extensive individualized planning and less able to actively participate in the planning of their own life content and maintain their personal autonomy (see Chapter 16 on interdependence). The family and service providers will have to make the most important decisions and assume planning responsibility.

Educational planning is concerned largely with preparation of appropriate content and organization. Making plans for the educational course of a person with JNCL requires comprehensive knowledge about the typical course of the disease, the individual variation (see Chapter 2) and the person's developmental history and current functioning. Selecting goals and making adaptations require thorough knowledge about the person's strengths and weaknesses, needs, interests and wishes. Planning should be based on assessment of all relevant domains (see Chapter 10). Development within each of these domains may follow its own course and may vary among individuals with JNCL. Each domain should be assessed separately: the person's functioning in one domain cannot be used to make assumptions about the person's functioning in other domains. For example, speech problems should not be used as an indicator of language comprehension or cognitive functioning. However, consideration of the person's needs must account for all domains individually and collectively. The educational plan will include activities that optimize the individual goals and interventions that ensure the necessary compensations to achieve the selected goals (see Chapters 2 and 12). Predicting the exact outcome of the interventions is never possible, and although plans should be based on a long-term aim, there may be frequent revisions of the plans and the goals that have been selected.

Person-centered planning

Students with typical development adapt and make their choices in accordance with the standard educational system. This implies that they have to adapt to *system-centered* plans, and the educational challenge is how to facilitate this process. Students with severe and complex disabilities are unable to adapt to the standard system, they need *person-centered* educational measures and interventions, a system that adapts to their needs (Meadan, Shelden, Appel, & DeGrazia, 2010; O'Brien & O'Brien, 1999). This in turn requires a person-centered approach. Person-centered planning has a different goal than standard planning. The basic question is what the system or society can do for the individual with a disability or disorder to obtain best possible learning outcome and secure the individual's rights and possibilities. A person-centered plan should be *proactive* and *realistic* in form. Choice of personal goals and interventions should lead to an optimal outcome within a realistic framework. The plan should optimize current and future possibilities of development and growth and be of help to avoid unnecessary problems and barriers at present and in the future. This might lead to some strain on the existing system, due to the needs for an individual-oriented organization, special competence or other resources, which might imply extra-ordinary costs.

Person-centered planning requires close collaboration between different agents who together have the knowledge required for making an optimal plan. Person-centered planning requires detailed knowledge about the person, which means that the family – and the person himself to the extent possible – will have a leading role in the planning process. Together, the family and the professionals representing the different agencies constitute a planning or responsibility group. It is a group rather than a team because parents are part of it. Here, it is called *a Responsibility Group* because the main task of the group is to allocate responsibility for the different areas to agencies or persons, thereby ensuring that it is clear who should do each of the tasks that have been decided by the group. If successful, the person-centered planning process will promote team building and cooperation. When the plan is completed, all the members should know their tasks and responsibilities.

In the present context, there are two types of person-centered plans that are relevant for people with JNCL: habilitation plans and education plans. Habilitation plans are usually comprehensive and include many life domains with a long-term perspective, often several years. Educational plans concern education and school life in a broad sense and usually have a one-year time perspective.

Habilitation Plans

The long-term plans for people with disabilities have been given different names, such as the Habilitation Plan, the Rehabilitation Plan, the Individual Plan or Individual Care Plan. In this context, the term Habilitation Plan will be used. The Habilitation Plan is mainly intended for people who have complex and comprehensive needs requiring support and adaptation in many life domains. The use of the Habilitation Plan is recommended by the authorities in many countries, and is a legal right in some countries, like in Norway where the habilitation plan currently is named Individual Plan (Norwegian Directorate of Health, 2018):

Anyone in need of long-term and coordinated health and care services is entitled to have an Individual Plan. The Plan should be prepared on the request from the person or his/her guardians. The local government service authority is responsible for developing the Plan.

The Individual Education Plan may be an integrated element of the Habilitation Plan. However, the Habilitation Plan is a holistic plan that goes beyond education, covering all relevant aspects of the individual's life related to social, educational, behavioral, recreational, residential, vocational, and medical needs. It is a written

plan of needs, interventions and actions, which outlines the goals and objectives selected by the individual, the family or the Responsibility Group, as well as the adaptations and compensations needed to achieve these goals and objectives (Baltes, 1997).

The Habilitation Plan may build on a positive and realizable vision of the person's functioning and achievements some years into the future, constructed and agreed upon by the members of the Responsibility Group. The Habilitation Plan lists the actions needed to fulfil the future vision described in the plan. These may include more short-term and detailed plans for the coming year (Hesselberg & von Tetzchner, 2008; Jeglinsky, Brogren Carlberg, & Autti-Rämö, 2014; King & Meyer, 2005).

The Responsibility Group should meet on a regular basis to evaluate existing goals and interventions, revise, reformulate or abandon long-term goals and decide on necessary changes to adaptations and interventions. An important function of the Habilitation Plan is to coordinate intervention efforts and ensure collaboration between different parties. The Habilitation Plan is person-centered and should function to empower the person and the family and ensure that the interests of the person are in the center of the discussion when goals are defined and selected.

The main purpose of the legislation related to the Individual Plan in Norway is to ensure that persons with social, psychosocial or medical needs are offered coordinated habilitation and rehabilitation services that can promote health, functioning, coping, participation and equality. Another purpose is to contribute to strengthen the interaction between service providers, service receivers and relatives. According to the authorities, the individual's own wishes should be emphasized. However, the personal goals and interventions described in the Individual Plan are not legally binding for the authorities.

The effect of a Habilitation Plan depends on the implementation of the measures that have been agreed upon. Habilitation Plans are sometimes criticized for being a mere formality with no real impact on the person's services. In spite of good intentions, the plan may be put in a drawer and forgotten, or the goals may be lost in the stress of managing everyday routine tasks. In the present study (Appendix A), some parents found that the habilitation plans functioned more as directives from the authorities than as a tool to promote this individual's goals:

The authorities have prepared a standard outline of the plan with significant limitations regarding content. It makes the plan more or less useless for us, the plan does not cater for our daughter's comprehensive needs. The plan is not the voice of our daughter, it is more a directive or overview of what we can or cannot expect from the society today and in future.

Actually, this quotation shows a violation of the intentions behind habilitation plans. The Habilitation Plan is supposed to be an overall plan to promote the best possible development, life situation and services for the person. It should include measures for making physical adaptations in the new environment, building necessary staff competence, and ensuring continuation in activities that are significant for the person and the social network. A Habilitation Plan should always have a calendar function for describing planned and achieved actions on a time line, when an initiative was going to take place, who was responsible for initiating and coordinating the initiatives or actions, and when the goals were achieved or revised.

Planning transitions

A transition may be defined as an important change in education, school setting or other life domains. The Habilitation Plan is an essential tool for planning transitions, for example from preschool to primary school, from primary to secondary school, or from attending school to attending a vocational activity center in emergent adulthood.

In some countries, students with JNCL are attending mainstream schools through all grades, although they may spend an increasing part of their time in smaller groups or special units. For students with JNCL attending special schools, there may be fewer transitions, as many special schools cover the full or most of the educational course. Students in mainstream schools therefore tend to be more affected by transitions than students in special schools. In some countries, students with JNCL first attend mainstream schools, then change to special schools for students with visual impairment, and finally move to schools for students with comprehensive needs (see Chapter 9). These transitions usually involve changes of teachers, staff, locations, classmates and sometimes changes of boarding facilities.

Some transitions may imply new rights or withdrawal of rights, for instance rights related to special needs education. The transition from school to not being part of an educational system in emergent adulthood (see Chapter 2) is a major life event for all persons with JNCL, independent of country and educational system. Education is a major structural element in all young people's life and usually has an even more central role for young persons with JNCL. In a successful transition, the role of the school must be replaced with sheltered workshops or similar, and planning and implementing this change is an important task for the Responsibility Group (see Chapter 23).

For students with typical development, a transition may be a positive experience, with new friends, new challenges, new teachers and maybe new opinions of the student's proficiencies. The situation may also be positive for

students with disabilities, but for many students with learning problems, and for those with JNCL in particular, a transition can be challenging. The competence of the individual and the staff's knowledge of goals and future needs may have to be rebuilt. Planning of transitions is therefore essential. Transitions are complex and if not thoroughly prepared, they may end in chaos and despair. The *educational flow* (see Chapter 16) may stop if competence and necessary compensatory equipment are not in place when students move to a new school. Students may experience a long time with restricted learning opportunities and loss of meaningful activities. Transitions should be thoroughly prepared and described in the Habilitation Plan because this plan has a long-term perspective. However, a separate short-term plan of action for the transition itself is often needed. The main objective of such a plan is to ensure continuity in learning and functioning, and to prepare the next school or service providers for meeting the individual with special needs in best possible manner (Elmerskog & Fosse, 2012). Moreover, transitions may involve domains other than education, such as health, social participation and activities outside school. A Habilitation Plan can also include needs that are not likely to be relevant in the short term but will be important in the future. For this reason, a Habilitation Plan should cover a period beyond the next transition.

Individual Education Plans

The Individual Education Plan (IEP) is a tool for planning and implementing education. An IEP is needed when the mainstream curriculum is not sufficient to meet the individual's educational needs (Mitchell, Morton, & Hornby, 2010). It typically has a time horizon of one year, meaning that it should be evaluated and revised at least once every year. However, the IEP should be in line with the goals and objectives described in the long-term Habilitation Plan.

The IEP is person-centered as it defines the educational needs and priorities for a single student (Keyes & Owens-Johnson, 2003). Students with learning disorders and disabilities are candidates for an IEP and many countries require an IEP when a student needs special education services. The IEP should address the student's learning challenges and needs, strengths and weaknesses, and include a specification of educational goals. It is a working document that may identify teaching goals and methods that are modified from the age-appropriate curriculum, or that represents an alternative educational content. It is a tool for the student, the parents, the school and all who are supporting the student to achieve the defined goals. An IEP should have clear and detailed descriptions of short-term goals, objectives, methods and resources needed. The right to have an IEP is legally binding in most western countries. It is strongly recommended

that parents are involved in preparing the IEP (Andreasson & Carlsson, 2013; MacLeod, Causton, Radel, & Radel, 2017).

Selection of goals is a core element in Individual Education Plans. The plans describe areas that should be given priority and reflect selection, optimization and compensation in relation to the student's goals and development (see Chapters 2 and 12). In the IEP, selecting goals also means selecting educational activities. For example, when tactile learning becomes important as vision deteriorates, the IEP should identify activities where the student can use the hands for exploration and creation. This is a characteristic of IEPs for students with JNCL. The plans include activities and learning based on anticipated declines that do not seem relevant at the time of planning but are important for the future.

The goals specified in the IEP should be proactive and optimistic, but also realistic, achievable and concrete (Elmerskog & Fosse, 2012; Pretti-Frontczak & Bricker, 2000). The educational goals should be specifically described because general formulations can make it difficult for teachers to decide which activities will most likely promote the selected goals. At the same time, the IEP should have a flexible content that can be evaluated and adjusted through the year. Evaluations may imply re-selection, adjustments and compensation.

Individual Education Plans for students with JNCL

Like the Habilitation Plan, the IEP should be based on assessments, knowledge about the disorder or disability and knowledge of the student. It should contain specified cognitive and physical goals and activities, which will give the student appropriate challenges. Experiences for example from New Zealand and Norway suggest that the IEPs for students with JNCL sometimes have a large proportion of goals related to entertainment, enjoyment and rest, and fewer related to the promotion of learning and development (Elmerskog & Fosse, 2012; Williams, 2008).

The activities chosen to realize goals should be based upon considerations that students with JNCL with increasing age will need more time and support to perform an activity or achieve a goal. Important issues for this group are what is necessary for new learning or for maintaining knowledge and skills, how the knowledge is going to be used at present and in the future, and how the students can use their knowledge and skills in everyday life outside school. For example, as the disease progresses, academic skills will gradually become a smaller part of their curriculum, while activities related to maintaining and managing activities in everyday life, such as going to the canteen, using the swimming-pool or cooking simple food, will take a larger part of the student's school time (van Delden, 2009). Students should be helped to remain independent as long as possible, even if the

time needed to accomplish the tasks increases. The declines will however sooner or later necessitate that autonomy to a greater extent is maintained through *interdependence*, where activities are performed together with another person (see Chapter 16). Learning by participation is therefore typically emphasized in the IEP of students with JNCL. There is a continuous process of learning to cope in new ways and with gradually more help.

The IEP will always include directions for the structure and organization of the teaching and the arrangements and adaptations of the classroom and other settings in school. The description of the organizational structure should extend beyond the student's classroom and include all parts of the school that are relevant for the student's education and social life. The educational measures should aim to ensure social affiliation, motor and cognitive learning and stimulation. Students with JNCL in mainstream schools will over time fall behind their peers in most subjects and may spend more time in a smaller classroom with a few other students with disabilities. This classroom should have a central location in the school and near the rest of the mainstream class to facilitate interaction with classmates and other peers.

An important function of the IEP is to contribute to maintaining appropriate expectations to the student. Expectations should neither be too high nor too low. The IEP should be written by teachers and other professionals who know the student with JNCL well in collaboration with the parents. In the present study (see Appendix A) many parents found the expectations to the student's educational possibilities too low. This might be caused by fears of failure in reaching educational goals or by an exaggerated focus on medical issues where the fatal and inevitable nature of the disease tends to overshadow educational possibilities. The IEP should state explicitly that it is neither harmful for the student, nor a reflection of failure, if a reasonable learning goal is not achieved as anticipated. However, to omit a learning goal or action, that could be attained if training were provided, can prevent students from developing their potential and reduce the zone of developmental maintenance (see Chapters 2 and 5). The present study indicates that many students with JNCL achieved learning goals far beyond what was anticipated in the beginning (see Chapters 12 and 14).

An IEP that focuses on what the student is unable to do rather than the educational possibilities may pose a severe threat to a student with JNCL. The IEP should express a clear belief that educational participation is important for development, activity, learning, cognitive maintenance and stimulation. Maintaining educational and social participation should always be essential elements in IEPs for children and adolescents with JNCL. The participation perspective should be explicitly formulated in IEPs for students with JNCL.

Reflections on educational planning

In the present study (see Appendix A), parents and staff were asked about the participants' IEPs. The IEP was more common than a Habilitation Plan, probably due to the fact that an IEP is legally required in many of the project countries. When the parents (N=107) were asked about their child's age when receiving an IEP for the first time, 72.9 percent specified the age (mean age 8.3, SD 2.4, range 4–16). Fourteen percent answered that they did not know, while 13.1 percent answered that their child had never had an IEP (N=69). The staff informants were asked only if the student had an IEP and according to them, 82.6 percent had an IEP, 8.7 percent did not have an IEP, while 8.7 percent of the staff said they did not know.

Among the parents whose child did have an IEP, 26.1 percent received the IEP before the school year started, 47.7 percent received the IEP after the school year had started, while 5.7 percent answered that the IEP was finished so late that it had no consequence for the education (Table 11.1). Parents were asked if the IEP was ready for use prior to the start of the new school year, but most IEPs were not ready at that time, partly due to the staff finding the planning difficult because of the nature of the disease or for bureaucratic reasons.

We as the parents have always requested the IEP to be ready before the beginning of the school year, the new IEP should be developed at the end of last school year... We would always ask the first week of school (August) and would generally get one mid to late October. The school considered it too much paperwork to do a full IEP at the right time.

The IEP was always late in secondary school, the IEP had no practical consequences for our child.

Table 11.1. Time of completion of the Individual Education Plan (IEP), according to parents whose children had received an IEP, and staff

	Parents (N=88) %	Staff (N=61) %
In due time before the new school year started	26.1	18.0
After the person had started the new school year	47.7	62.3
Too late, no practical consequences for education	5.7	1.6
Don't know	20.5	11.5
No IEP	-	6.6

The first IEP was way too late – it took about 1½ years from the learning and educational problems were identified until we had an IEP.

Parents and teachers emphasized a need for support from JNCL specialists when developing an IEP:

IEPs are so important for us parents. It must be a written document, and followed up in collaboration between teachers, parents and JNCL supervisors.

The work with IEP and the collaboration became good when specialists from the School for the Visually Impaired in Jyväskylä were involved.

JNCL specialists and parents are important contributors of IEPs, it is wrong to expect teachers with minimal knowledge about JNCL to do this important work without guidance from supervisors with sufficient knowledge.

In addition, 20.5 percent of the parents did not know when the child's IEP was completed, indicating that they did not participate in the work with the education plan. The staff informants were asked the same questions as parents (Table 11.1). The fact that 11.5 percent of them did not know if the student had an IEP suggests that the staff who were interviewed had not been engaged in the work with the IEP.

Several parents were critical to how the IEP was handled by the educational authorities:

The IEP was denied by the educational authorities in the municipality, my child has not been given an IEP so far.

The local authority (Primary Care Trust as it was then) took limited interest most of the time, no IEP.

We wanted an IEP for our child, but I felt our local school district didn't want us around and they tried very hard not to work with us, we had to fight for everything we wanted.

Comments from some staff members indicated that they found it difficult to plan for the student with JNCL. Some even considered all planning unnecessary or unethical for students with JNCL because of the course of the disease, which is apparent in the following quotations:

This process with the Individual Educational Plan has been changing and it is difficult to comment on the impact of the IEP on something that is evolving.

I sometimes feel the work done with IEP is wasted time. Suddenly the condition is so different.

I am not sure it is fair to make plans when you know the outcome of the disease.

It feels wrong to make plans for the future when nobody knows what is going to happen.

IEP is not possible because of the disease.

IEP for this school year is delayed because of the child's illness.

Teachers and other staff members also underlined that the IEP had room for modifications and revisions when needed. However, there was no evidence that education and IEPs were based on the habilitation plan. Educational measures seemed mainly to be based on the current situation. There was little or no attention to the progression of dementia and the need for precautionary or hastened learning and educational life flow (see Chapter 12). These concepts imply a need for «planning for tomorrow's needs» (proactive learning and teaching), and a need to teach to an automated level of mastery so that ultimately, the skill thus taught can be executed with minimum demands on cognitive monitoring.

Parents and educational staff were asked about the impact of the IEP on the students' learning, development and maintenance of skills. The results showed that 61.2 percent of the parents and 53.9 percent of the staff evaluated the impact to be high or very high, while 15.7 percent of the parents and 23.1 percent of the staff found the impact moderate, and 11.2 percent of the parents and 4.6 percent of the staff found little or no impact. The parents and the staff thus had quite similar evaluations of the impact of the IEP.

Parents and staff were asked about the parents' role in the development of the IEP. Forty-four percent of the parents and 33 percent of the staff answered that the parents had been important contributors to the IEP. Most countries emphasize collaboration with parents in the development of the IEP (MacLeod et al., 2017). However, the parents' comments about IEP and the relatively low degree of parent contribution indicate that the current practice needs improvement. In fact, several parents suggested that the IEP was not used primarily for achieving the best possible education for their child but rather to achieve other goals:

Table 11.2 Impact of the IEP according to parents whose children had received an IEP, and staff

	Parents (N=89) %	Staff (N=65) %
No impact	2.3	0.0
Low impact	8.9	4.6
Moderate impact	15.7	23.1
High impact	29.2	27.7
Very high impact	32.6	26.2
Don't know	11.2	10.8
No IEP	-	7.7

I am sitting with a feeling that the IEP was done to satisfy the authorities, not the needs of my child.

The educational authorities demand that IEP is written according to the needs of the authorities. We feel we use a lot of time on paperwork to satisfy the authorities instead of our child.

Some parents found the IEP too vague, it was not really possible to find out what would be happening in school, which the following quotations indicate:

I do regret that we were not specific enough about the amount of time and sessions for the music therapy, speech therapy and hydrotherapy.

The IEPs were vague in the targets but vital to ensure that resources are in place and funded.

The activities defined in the IEP were not always implemented:

When BDSRA came out and helped with our latest IEP a suggestion she had was to implement objects that would be significant to our child for transitions (a steering wheel to go to the bus, another object when it was time to go to specials, and so forth). To date, we have not heard about them implementing this and his IEP was completed in September.

However, most parents and staff expressed the view that educational planning is important for students with JNCL and has a positive impact on learning, development and maintenance of skills.

Parents (N=112) were asked to appraise the information they had received from different contributors during the first year after the diagnosis was confirmed on a scale from 1 (no contribution) to 5 (very high degree of contribution). Parent organizations received the highest mean score (3.6), followed by other parents in same situation (3.4) and competence centers (3.3). The local educational authorities got the lowest mean score (1.4). These results document the need to include JNCL specialists early in educational and other planning.

The staff was asked to appraise the role of different contributors to their own competence building related to visual impairment and JNCL. Collaboration with parents got the highest mean scores (3.8 for visual impairment and 3.7 for JNCL), followed by competence centers (3.2 for both visual impairment and JNCL). The local educational authorities got the lowest mean scores (2.1 and 2.0).

These results probably reflect that JNCL is a very rare disease and that parents, teachers and other staff did not find sufficient competence in the local services. As part of the present project, a literature search was performed, and the result was almost exclusively articles from the medical domain. This indicates that JNCL and childhood dementia are practically unknown in both the educational and the social services. The absence of adequate knowledge may lead to lack of initiative, disillusionment and discouragement, because professionals become likely to focus mainly on the negative medical aspects of the disease and not on the educational possibilities and interventions. It is noteworthy that information from the parent organizations (see Chapter 24) was considered important by many parents, emphasizing the importance of having contact with other parents, for both the educational planning for their own child and for supporting other parents.

Parents were further asked about the impact learning during school years had had on adult living, on a scale from 1 (no impact) to 5 (very high impact). The two school subjects with the highest average were physiotherapy (4.1) and Music/music therapy (3.9), followed by physical education/training (3.5) and social gathering with other students (3.5). The lowest scores were for mathematics (1.9) and nature studies (2.1) but even if these domains had less apparent impact, they represent general knowledge in society and participating in such classes is part of inclusion.

Principles for making plans for persons with JNCL

Educational planning for persons with JNCL differs from planning for most groups who need special education. Plans usually focus on growth in knowledge and skills, on identifying the zone of proximal development and learning potential, and appropriate adaptations and educational strategies (see Chapters 2 and 12).

This is also important for individuals with JNCL but in addition, planning must take the possible emerging declines and losses into account. Planning should include adaptations and educational strategies in areas with independent and interdependent functions within the zone of developmental maintenance (ZDM). Figure 11.1 shows learning and development in two hypothetical persons with JNCL, who have different areas of strength and weakness, and hence need different interventions. The figure illustrates that functions may be maintained to different degrees when independent function is reduced, but maintenance depends on appropriate help and support.

The standard planning procedures that are used for students in general should be utilized as far as possible for students with JNCL, meaning that they share goals, activities and themes with their peers in an inclusive setting. However, because students with JNCL require new educational measures as the disease progresses, there will be a need for planning for lower functioning in the future. Interventions that build on growth are within the zone of proximal development, while interventions preparing for or following decline are within the zone of developmental maintenance, applying Baltes' (1997) principles of selection (choice of relevant and achievable goals), optimization (forming and maintaining means to achieve the goals) and compensation (new strategies to maintain functions and goals that are no longer sustainable in ordinary ways) (see Chapters 2 and 12). This will include developing an extended curriculum and interventions in areas of special importance for each student. This will usually involve areas and strategies that ordinary teachers and other staff in mainstream educational settings are not familiar with and should therefore be emphasized in the IEP and the Habilitation Plan.

The «The Participation and Performance Dilemma» (Gray & Hollingsworth, 1999) is illustrated in Table 11.3, adapted to the education of persons with JNCL. The table shows four possible scenarios: In Scenario 1, the individual is performing and participating in an activity. This scenario represents the general situation in education. In Scenario 2, the individual is not performing but still participating in the activity. This scenario is rather uncommon since education typically is based on both participation and the student's performance in the activity. However, Scenario 2 may represent a goal for individuals with JNCL. The declines may make performance within the activity difficult, while assistance, adaptations and interdependence may make participation and engagement possible, for example in mathematics or art (see Chapters 12 and 16). Scenario 3 does not represent a desirable situation and may be a result of poor planning. For instance, the person has learned to use braille but is not provided with materials that enable him to use his tactile reading skills, because the staff at the person's new residence is not familiar with braille and not aware of the person's abilities. In Scenario 4,

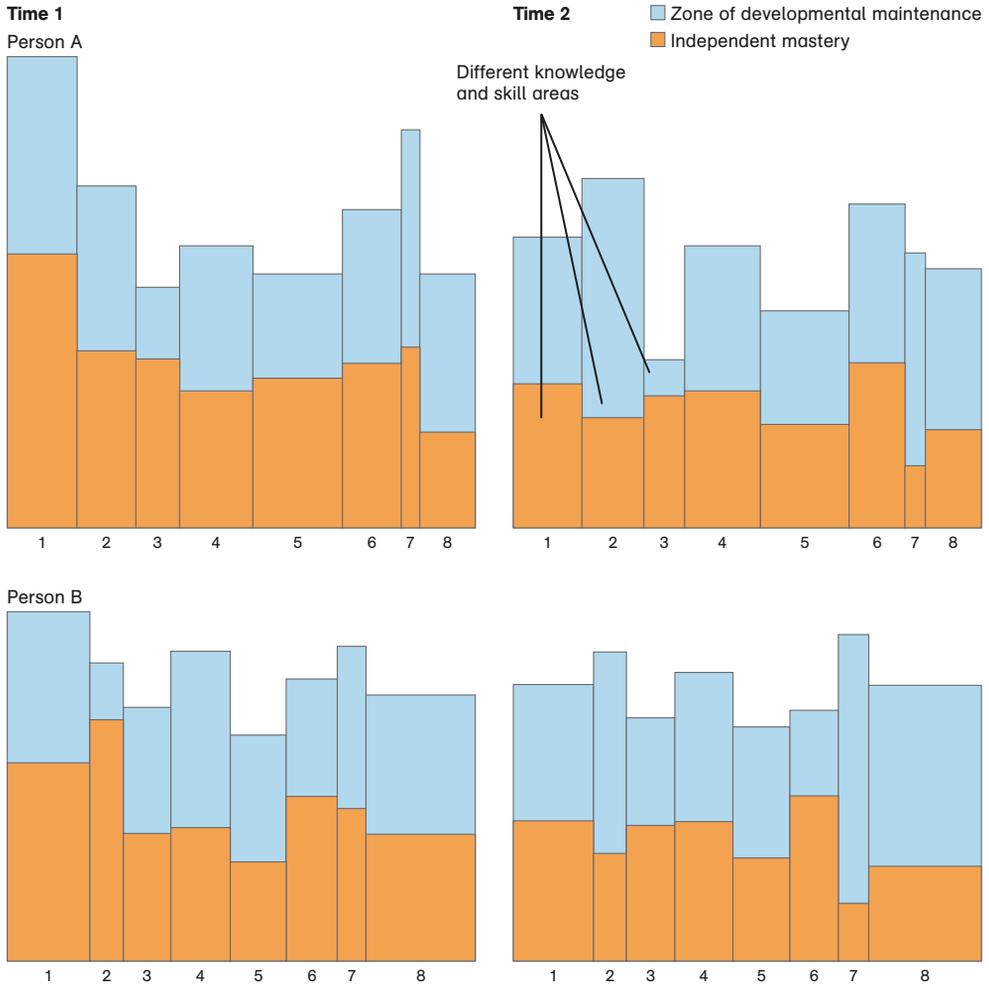


Figure 11.1 Zone of Developmental Maintenance (ZDM)

The figure shows independent performance and the zone of developmental maintenance (what a person is able to do with help) for two persons at two points in time, and for eight knowledge and skill domains. The persons differ in knowledge and skills and the differences between the domains are shown as variation in "width" and "length". The developmental courses of the two persons differ also somewhat from Time 1 to Time 2. In domain 1, both independent performance and ZDM have decreased from Time 1 to Time 2 in the two persons, although somewhat differently. In domain 2, independent performance has decreased and the ZDM increased correspondingly in both persons. They have lost independence but have maintained the function with help (interdependence). In domain 4, the ZDM of Person B has decreased (can do less with help), but there is no change in the person's independent performance. For Person A neither independent performance nor ZDM have changed in this domain. In domain 7, independence was reduced in both persons from Time 1 to Time 2, but the ZDM remained the same in person A and increased in person B. Consequently, the two persons have different strong and weak domains and show different developmental declines between Time 1 and Time 2.

Table 11.3: The Participation and Performance Dilemma (based on Gray & Hollingsworth, 1999)

	Ability to do activity	Inability to do activity
Participation	Scenario 1 Assessment shows Performance and Participation	Scenario 2 Assessment shows No Performance but Participation
No participation	Scenario 3 Assessment shows Performance but No Participation	Scenario 4 Assessment shows No Performance and No Participation

the activity has been terminated because it was now outside the person's zone of developmental maintenance and thus not relevant for the person anymore. However, Scenario 4 may also be a result of poor planning and too little support.

The scenarios in Table 11.3 raise several questions. If a situation is described as Scenario 2 or 3, one may ask what initiatives are needed to achieve Scenario 1 (if desired and feasible). If the situation is described as Scenario 4, one may ask what initiatives are needed to achieve Scenario 1 or 2 (if desired and feasible). There is always a risk that low performance will lead to the assumption that the activity is not suitable for the person, and subsequently to non-participation. Participation must therefore be the core element in planning for maintaining or improving skills and quality of life in persons with JNCL, also when independent performance is limited. Participation is about personal engagement and should not depend on the ability to perform. An important objective in the planning work is to promote initiatives that will allow persons with JNCL to participate in desirable activities and provide adequate support and adaptations to make participation possible.

For persons with JNCL, participation may depend on proactive planning. In the later stages of the disease, persons with JNCL may need extensive support from helpers. The need for individual support in all daily activities should be described explicitly in the plan. Preservation of personal engagement, participation, skills and quality of life all depend on appropriate support. The guiding questions of planning will thus be: What can the person with JNCL do and participate in when supported by a helper, and what help and support are necessary for maintaining performance and participation?

Core issues in educational planning

The educational system is based on expectations of acquisition of knowledge, skills and attitudes in predefined domains. The student is expected to achieve defined goals, for instance within literacy and mathematics in accordance with a

curriculum. For students with JNCL, the processes of planning and implementing educational interventions will undergo comprehensive changes during the individual's life time (see Chapter 12).

Participation in educational and social situations is important for students' learning and quality of life (Centre for International Development and Training, 2001). No curriculum can be compared to what children and young people learn through participation in everyday situations and special events, that is, by being engaged in situations where things happen. Participation-based learning is an implicit general goal in society where participation is taken for granted (United Nations, 2018a,b). It is usually assumed that participation eventually will be maintained by the individual. Participation-based learning facilitates implicit learning, that is, the acquisition of knowledge and skills that occur incidentally, without planning and without personal awareness of the fact that learning is taking place (Frensch & Runger, 2003). Planning for participation in everyday activities is therefore a way to promote implicit learning and development in students with JNCL in all phases of the disease. Students with JNCL may to some extent be able to ensure participation in the early school years but will need help to participate in activities with implicit learning when explicit learning becomes more difficult to achieve. Possibilities for participation-based learning should be included in IEPs of all students with JNCL and become more emphasized with age. Planning to promote participation is therefore about creating a life agenda for the student, with specified meaningful activities from early morning to bedtime, based on knowledge about the student's interests and possibilities. Maintaining and securing educational and social participation through participation-based learning is a central element of interventions for students with JNCL, irrespective of how they function.

Precautionary or proactive teaching and learning mean the teaching of skills that the student does not need at present but will need in the future (see Chapter 12). Teaching augmentative and alternative communication when the student's speech is still intelligible is an example of early learning to later problems, in this case when speech is no longer intelligible to others (Chapter 13). Early learning of how to operate a computer and software programs adapted for individuals without vision will prepare the student for a future loss of vision. However, it is unusual in special education to prepare for future losses of skills. Some of the parents and teachers in the present project described resistance to proactive teaching activities. Some teachers thought it was unethical to address a problem which was not yet present (see quotations from staff and parents, Chapter 14). There were also doubts about the students' motivation to learn something when they were unaware of their need for it. Proactive learning was perceived as somewhat odd, in particular when the student was not aware of the diagnosis. However, the project also found

good examples of proactive teaching and learning in students with JNCL (see Chapters 13, 14 and 16). It depended on the enthusiasm and dedication of the people in the planning group, and on how the teaching activities were organized. The ethical dilemma related to proactive teaching is illustrated by the following statement, based on many quotations from parents in the current project: *It was too late to start with the learning program, the cognitive decline had gone too far* (see also Chapter 9). In *hastened learning*, intensified teaching is planned to accelerate the student's learning in selected and complex areas which require best possible cognition, when this is considered advantageous in the longer term (see Chapter 12).

Maintaining *life flow* means to have a certain degree of continuity (see Chapter 16). This principle is of special importance for planning of transitions, which often can threaten the life flow of the individual. A dementia perspective acknowledges that the person's history and experiences, as well as skills, interests and social life should be addressed in the IEP and Habilitation Plan. The major transitions may be experienced as dramatic, and if not considered in the plans, may create chaos in the person's life and negatively influence the person's mental health (see Chapter 27). The JNCL project has disclosed examples of severe breaches of the individuals' life flow in connection with transitions.

The principle of life flow is always important in planning for people with JNCL but will become more decisive with age. Transitions should be planned and prepared in close collaboration between the family, the person when possible and all relevant service providers. However, this is not always the case, as evident in the following quotations from parents:

When he transitioned from elementary school to middle school, the person in charge of special education recommended he be placed in a classroom that focused on daily living skills as opposed to regular subjects like math and science. This was actually done without our knowledge. He was very bored.

We as parents always had to organize the transitions on our own. We never received any support.

The transition to adult living was difficult, we had to accept what was there, the needs of our son was not important.

The transition to adult living is further discussed in Chapter 23.

A planning tool for JNCL

Children and young people with JNCL should have similar plans, educational measures and life contents as their peers when this is possible and to their benefit. Participation in ordinary activities and sharing of knowledge and goals with others will promote social inclusion and participation in society. However, children and young people with JNCL need more than the standard plans. The unique and different perspectives on planning for students with JNCL are anticipation of future challenges and consideration of possibilities despite these challenges. Anticipating future challenges and implementing measures to meet these challenges can be a very emotional process, especially for parents but also for staff. Such issues must be handled with great respect and empathy by all involved. Professionals should all the same be able to make plans for future challenges and possibilities and act accordingly with or without parent participation. Assessment is necessary for planning to be based on the best possible knowledge. The Educational Development Observation tool (EDO) is designed to give an overview of themes that should always be considered in planning for persons with JNCL and thus constitutes a knowledge foundation for the IEP and the Habilitation Plan. The main themes in the EDO are vision, communication, literacy, social life, gross motor function, fine motor function, physical activity, behavior, memory and attention, independence and autonomy, interests and equipment (see Chapter 10). From a long-term perspective, the EDO is making it possible to address domains, which will decline sooner or later. From a short-perspective horizon, the EDO helps see what can be done at present to meet the future challenges. It is particularly important to use the EDO prior to major transitions, such as transitions between schools, or the transition into adult living.

Conclusions

Making plans for a child, adolescent or adult with JNCL requires knowledge about JNCL and the individual. The overall needs are comprehensive and wide-ranging, and it is necessary to involve JNCL specialists and parents in the planning of education and other interventions. Observations and discussions must be initiated at the right time according to a timetable based on an anticipated progression of dementia. The ongoing or anticipated symptoms caused by the disease should represent a working agenda for the Responsibility Group. What is not done today may be difficult to achieve tomorrow. The development of dementia sets the framework for educational planning and interventions. Education cannot stop the disease from progressing, but education can meet the symptoms with selection, optimization and compensation measures.

References

- Andreasson, I., & Carlsson, M. A. (2013). Individual Educational Plans in Swedish schools – forming identity and governing functions in pupils' documentation. *International Journal of Special Education*, 28, 58–67.
- Ballantine, J. H., Hammack, F. M., & Stuber, J. (2017). *The sociology of education: A systematic analysis*. New York: Routledge.
- Baltes, P. B. (1997). On the incomplete architecture of human ontogeny: Selection, optimization, and compensation as foundation of developmental theory. *American Psychologist*, 52, 366–380.
- Centre for International Development and Training, CIDT (2001). *Participation, learning and action (course handbook)*. Walsall, UK: University of Wolverhampton.
- Elmerskog, B., & Fosse, P. (2012). *Lære for livet. En innføring i pedagogikk og læring ved juvenil neuronal ceroid lipofuscinose (JNCL)* (Learning for life. An introduction to education and learning in juvenile neuronal ceroid lipofuscinosis (JNCL)). Trondheim: Akademika forlag.
- Frensch, P. A., & Runger, D. (2003). Implicit learning. *Current Directions in Psychological Science*, 12, 13–18.
- Gray, D. B., & Hollingsworth, H. (1999). Development of new measurement tools for studying participation in context. Presented at American Public Health Association Annual Meeting, Chicago, Illinois, November 1999.
- Hesselberg, E., & von Tetzchner, S. (2008). Planlegging av framtiden (Planning for the future). In S. von Tetzchner, E. Hesselberg & H. Schiørbeck (Eds.), *Habilitering – Tverrfaglig arbeid for mennesker med utviklingsmessige funksjonshemninger* (Habilitation – multidisciplinary work for people with developmental disabilities) (pp. 64–84) Oslo: Gyldendal Akademisk.
- Hesselberg, E., & von Tetzchner, S. (2016). *Pedagogisk-psykologisk arbeid* (Pedagogical-psychological work). Oslo: Gyldendal Akademisk.
- Jeglinsky, I., Brogren Carlberg, E., & Autti-Rämö, I. (2014). How are actual needs recognized in the content and goals of written rehabilitation plans? *Disability and Rehabilitation*, 36, 441–451.
- Keyes, M. W., & Owens-Johnson, L. (2003). Developing person-centered IEPs. *Intervention in School and Clinic*, 38, 145–152.
- King, G., & Meyer, K. (2006). Service integration and co-ordination: A framework of approaches for the delivery of co-ordinated care to children with disabilities and their families. *Child: Care, Health and Development*, 32, 477–492.
- MacLeod, K., Causton, J. N., Radel, M., & Radel, P. (2017). Rethinking the Individualized Education Plan process: Voices from the other side of the table. *Disability and Society*, 32, 381–400.
- Meadan, H., Shelden, D. L., Appel, K., & DeGrazia, R. L. (2010). Developing a long-term vision: A road map for students' futures. *Teaching Exceptional Children*, 43, 8–14.
- Mitchell, D., Morton, M., & Hornby, G. (2010). *Review of the literature on individual education plans*. Christchurch, NZ: University of Canterbury.
- Norwegian Directorate of Health (2018). Veileder om rehabilitering, habilitering, individuell plan og koordinering (*Guide to rehabilitation, habilitation, Individual plan and Coordinator*). Retrieved from <https://helsedirektoratet.no/retningslinjer/rehabilitering-habilitering-individuell-plan-og-koordinator>
- O'Brien, C. L., & O'Brien, J. (2000). *The origins of person-centered planning: A community of practice perspective*. Lithonia, GA: Responsive Systems Associates.

- Pretti-Frontczak, K., & Bricker, D. (2000). Enhancing the quality of individualized education plan (IEP) goals and objectives. *Journal of Early Intervention, 23*, 92–105.
- United Nations (2018). Convention of the Rights of Persons with Disabilities. Retrieved from <https://www.un.org/development/desa/disabilities/convention-on-the-rights-of-persons-with-disabilities.html>
- United Nations (2018). Convention of the Rights of the Child. Retrieved from <https://www.ohchr.org/EN/ProfessionalInterest/Pages/CRC.aspx>
- van Delden, G. W. (2009). Education for children with Batten disease. Presented at 7th European Conference of ICEVI, Dublin, Ireland.
- Williams, L. A. (2008). Students first: A trans-disciplinary team approach to the education of a student with Batten Disease. Retrieved from Internet August 17th, 2018. https://ir.canterbury.ac.nz/bitstream/handle/10092/2647/Thesis_fulltext.pdf;sequence=1

12

Early and Later Educational Intervention

*Anne-Grethe Tøssebro, Stephen von Tetzchner,
Bengt Elmerskog and Aine Murphy*

The education of individuals with juvenile neuronal ceroid lipofuscinosis (JNCL) will depend on the competence in the educational system, as well as the society's general traditions in mainstream and special needs education. A search for research on planning and implementation of education for students with JNCL yielded very little, and there are hardly any studies about the impact and efficacy of educational practices for students in this group (von Tetzchner, Fosse, & Elmerskog, 2013; Williams, 2008). Educational systems differ a lot between countries, also among the countries participating in the present project (see Chapter 9). There are a few published guidelines for teachers about education for students with JNCL (Bills, 2011; Elmerskog & Fosse, 2012). In general, students with JNCL follow the same curriculum as peers for as long as possible. Some countries have special schools and institutions with considerable experience in working with students who have JNCL, in other countries students in this group attend mainstream schools with no prior experience in teaching a student with JNCL.

In recent years, two general principles have been emphasized in special education: selection of individual educational goals and individual adaptation. The teaching should meet individual needs and utilize individual strengths, and this applies to students with JNCL as to all other students.

Individuals with JNCL develop childhood dementia, which is dementia with onset in childhood (see Chapter 5). There are several small diagnostic groups with cognitive and other declines in childhood (Schoenberg & Scott, 2011) but the number of children is small and childhood dementia is a rather unknown concept in education, also in special schools and schools with children who have degenerative diseases. Compared to education and special education in general, students with childhood dementia require a significant change of practice, and the selection of educational goals and individual adaptations is partly based on different assumptions and premises.

The education of students with JNCL must be adapted to the present and future changes in their learning abilities, to what the students are able to do and learn in the present situation and in the near and distant future. The period before the onset of decline in a domain may be considered a «time window» or «window of opportunity» for the student to learn particular skills for later use. For instance, the educational strategies will change from utilizing residual vision to relying on auditory and tactile information, and educational strategies may be directed at precautionary learning (see below) to meet future needs (Elmerskog & Fosse, 2012).

All students need continuous changes in education to meet their knowledge, abilities and skills, mostly as a result of their intellectual growth. Students with JNCL need continuous educational adaptation to meet both growth and decline in their abilities. This means that teachers need knowledge about the functional consequences of the disease and the educational strategies that may support learning and coping (Elmerskog & Fosse, 2012; Uvebrandt, 2006). Because of the limited knowledge, there is a need for systematic experience and developing new strategies. The model proposed by Baltes and Baltes (1990) may be a useful tool for selecting goals, optimizing methods for reaching educational goals, and developing new ways to compensate for functions that are slowly deteriorating (see Chapter 2).

This chapter describes changes in practices through the educational course of students with JNCL, from a focus on academic subjects to more practical domains. Academic subjects may include mathematics, writing and reading, physics, native and foreign languages, geography, and history. Some require more reasoning skills while others require an ability to learn facts. Reasoning is usually difficult for students with cognitive problems. Remembering facts can be easier for students with JNCL, in particular when facts are related to an area they are interested in (Elmerskog & Fosse, 2012). Practical domains may include physical education, woodwork, home economics, domestic science, and orientation and mobility. These are often associated with how to do tasks and procedural learning (see Chapter 16).

Several chapters in this volume include examples of educational adaptation. At the end of the present chapter, the adaptation of two school subjects is described. Mathematics represents an academic subject while drama is a practical school subject, facilitating the achievement of personal, social and cultural identity.

Adaptive education and special needs education

Education for children and adolescents with JNCL usually includes both adaptive and special needs education. In *adaptive education* the ordinary curriculum is made accessible for students with special needs (Fasting, 2013). For instance, learning how to read the clock is a goal within the mainstream curriculum, and can be adapted to students with visual impairment by using a tactile or auditory clock, and by introducing quarters of an hour instead of minutes in the early intervention when needed. Similarly, braille or text-to-speech programs may make textbooks accessible for this group of students.

Simon was working with addition and subtraction, using an electronic workbook on his computer. The text in the workbook was read aloud with synthetic speech, and Simon wrote his answers into the book with a braille keyboard. These adaptations made it possible for Simon to work with the same material and having the same goals as his classmates.

Special needs education may be defined as the adoption of educational goals and associated strategies not usually applied with children of the same age group. In this definition, special needs education will always imply a change in curriculum. For instance, learning the mobility route from the classroom to the schoolyard would be a goal within special needs education for students who are blind because it is not part of the ordinary curriculum. A student with JNCL will not share this education goal or the training with sighted peers in a mainstream setting.

The distinction between adaptive education and special needs education is not always clear. For example, literacy is a main goal in both mainstream education and special needs education but different modalities and strategies are applied. Teaching of tactile reading and writing strategies is not part of the ordinary curriculum and sighted peers do not usually participate in the training needed to learn braille or Moon.

The child's rights and needs

In many countries, adaptive education and special needs education are legal rights in the educational legislation. Irrespective of the need for either of these rights, it is a commonly held view that education of students with typical development and students with disabilities should consider the students' strengths and limitations, as well as possibilities and barriers in the educational environment (Elmerskog, Storliløkken, & Tellevik, 2008). It is important to know the child, the social environment and the culture (Rogoff, Dahl, & Callanan, 2018).

Three educational phases

The education of students with JNCL can be divided into three main phases. The results of the present study (Appendix A) indicate that the average age when students with JNCL begin to show problems with learning and remembering new things is around 10–11 years, but also that the age of onset of cognitive and other declines vary considerably within the group (see Figure 1.1). The phases therefore do not follow age but the students' developmental declines and general functioning, and the length of each phase will vary.

The phase with no or mild dementia

At the time of diagnosis, cognitive problems of students with JNCL are usually not very pronounced and they may be able to follow the mainstream curriculum for some time (Elmerskog & Fosse, 2012; Uvebrandt, 2006). The present survey found that some of the skills and interests that the students develop early in life will last for a considerable length of time. This early phase is therefore important for building interests and skills that may be used in education and everyday life in the later phases of the disease.

Flexibility and variation

Participating in class during the early school years facilitates the exposure to ordinary school subjects and themes. Mainstream education implies flexibility and variation, which will benefit both the child with JNCL and the classmates. The teaching focuses on new knowledge and skills and switches between different subjects several times each day. This period of relatively intact cognitive functioning represents an important learning window for students with JNCL and is of high importance for capacity building and for establishing individual interests.

Adam is very interested in dinosaurs. He has a lot of knowledge about the different species. The interest was kindled early in life and has been an inspiration both for learning and remembering facts and developing writing skills. Every day, Adam is writing facts and stories about dinosaurs on his computer with a braille keyboard.

Most children with JNCL follow the ordinary curriculum as long as possible, acquire new knowledge and skills, and develop concentration and problem solving ability within many subjects and themes. Learning is to a large degree explicit, skill-oriented and focused on capacity building.

Learning through vision and visual memories

From an educational perspective, it is a huge advantage that students with JNCL have had normal eyesight in the first years of life. Reference to their visual memories allows them to think visually and they will associate early visual experiences with experiences based on tactile or auditory perception later in life. For example, prior visual experience facilitates students' early learning of mathematics.

School beginners with JNCL experience a gradual deterioration of visual functioning, but some students retain residual vision for several years (see Chapter 4). During the first years, children with JNCL can usually use visual learning strategies to some extent. They learn many daily living skills through visual observation, such as washing the hands or turning on the radio or television. Teachers must monitor the student's visual functioning carefully, in order to know when it is necessary to introduce strategies based on other modalities.

Learning in a social setting

Being a part of a class or a group and participating in the same educational and social activities as peers is highly motivating for most children. Students with JNCL will eventually need technical aids and special learning materials that make them appear a little different, such as magnifying devices with closed circuit television (CCTV) and equipment for reading and writing braille (see Chapter 14). Writing braille on a Perkins braille or on a braille display with synthetic speech feedback creates noise and may be a distraction for others in the classroom; therefore ways should be found to minimize distractions and facilitate full participation. The use of head-phones will reduce the noise but make it more difficult for the student to hear instructions from the teacher and other important sounds in the classroom. Such adaptations may interfere with inclusion goals and should only be used when necessary. It is important to organize the classroom and monitor the activities in a way that ensures the best possible working environment both for the student with JNCL and the classmates.

Susan had some residual vision and used CCTV to magnify texts and illustrations in the textbooks. She was dependent on this equipment to do her school work. However, she had never felt quite comfortable with these technical aids because she in this way became different from the classmates. To prepare for future needs it was time to introduce Susan to braille. Teachers were concerned about how she would react when becoming even more different by learning and practicing tactile reading. To «normalize» braille the whole class was introduced to braille reading and writing during a visit from the national competence center. The classmates read braille letters using vision. The introduction of braille became a success for Susan because of the enthusiasm the other students expressed.

Comments like «This is the most exciting thing I have learned for a long time!» from a classmate made a big difference for Susan's attitude to braille.

Reducing noise, making the environment physically inviting and allowing other students to learn to use the special equipment will contribute to inclusion and social affiliation for students with JNCL.

Precautionary learning

Precautionary or proactive learning implies that children are introduced to new skills they will need in the future but not in their present situation. The gradual cognitive decline in JNCL will make acquisition of complex skills increasingly difficult. Complex cognitive and tactile skills, like reading and writing braille or Moon, should therefore be introduced as early as possible, preferably very soon after the diagnosis is confirmed (see Chapter 14). The time window for learning tactile reading is in early childhood when the child's learning capacity is best. However, the present survey found that many students with JNCL were not introduced to braille before their visual impairment made visual reading impossible (see Chapter 14). By this time, the time window for learning braille might be closed because of the student's cognitive decline.

Some skills are pivotal in the sense that they establish a foundation for further learning and functioning, for example literacy skills and basic mathematics, and such skills should be automatized when possible. This automatization has been described within the concept of *hastened learning* (Tellevik, 2008, see below). Automatization means skills are mastered so well that they require little conscious effort. Automatization requires massed training and many repetitions during the learning period. Automatized skills are also resistant to decline because they require little cognitive capacity. Promotion of pivotal skills should therefore be given priority in the early education of children with JNCL.

Hastened learning

The principle of *hastened learning* concerns intensified instruction to accelerate learning in selected areas. The underlying assumption is that the student's current capacity enables learning of skills that might be difficult or impossible to learn in a later phase, such as writing and reading or playing the piano. Hastened learning may also be used to facilitate the development of early interests that might be of importance in later phases, such as music and sports (see Chapters 14, 16, 17 and 23). The aim of hastened learning is thus to achieve a level of performance that will require little cognitive capacity or strain when it has been acquired, and thus represent good cognitive economy (Tellevik, 2008). Hastened learning will usually require explicit teaching of skills.

Selection, optimization and compensation

For most students with JNCL the selection of goals in the Individual Education Plan (IEP) in this period should reflect the same range of subjects and themes as for their classmates. There are however, some additional domains that should be prioritized because of the visual deterioration, such as tactile reading and writing (see Chapter 14), mobility and orientation, and autonomy (see Chapter 16). The demand for special teaching can feel overwhelming to teachers responsible for implementing the training. In addition, teachers might perceive that the demands on the students' learning capacity exceed an acceptable level. When prioritizing is necessary, there should be a focus on knowledge and skills that will be useful for the student both at present and in the future (see Chapter 11).

In this process, Baltes' (1997) principles of selection (choice of relevant and achievable goals), optimization (forming and maintaining means to achieve the goals) and compensation (new strategies to maintain functions and goals that are no longer sustainable in ordinary ways) may be applied. Optimization for achieving the individual goals may imply new activities and strategies both for the teacher and for the student with JNCL. For example, staff may need to mediate visual information by giving the necessary information verbally and the student may have to learn to use technical equipment in order to read and write a tactile alphabet. Activities related to teaching route planning and mobility will ensure successful locomotion on the school premises and greater autonomy (see Chapter 16).

Like other students, students with JNCL learn best when the educational situation is exciting and positively challenging, and when the content is meaningful and matches their skills and interests (Norris & Closs, 1999). Ensuring a proactive and functional IEP for a child with JNCL requires the joint efforts of the school team as well as support from specialists with competence in visual impairment and JNCL (see Chapter 11).

The moderate dementia phase

As students with JNCL become older, the need for individual adaptation gradually increases. Their ability to maintain attention over time, listen to the teacher and remember spoken information decreases. The students benefit from teaching involving personal experiences and meaningful activities with sensory information from multiple sources, such as in the example below.

Lisa was a student in secondary school and attended an ordinary class. The teacher was telling the class about the different nutrients in dairy products and Lisa was struggling to keep attention and remember what the teacher

was saying. The teacher therefore chose another approach for Lisa. Together with Lisa, she prepared an interview with ten questions about dairy products and made an appointment with the local dairy for a visit and interviews with members of the staff. Lisa was given a tour around the dairy, where she could taste and smell some of the products and listen to the production sounds. Lisa was able to complete her task with an adult assistant. Learning became much easier for Lisa, she could work with the same educational goals as her classmates, but in her own way and speed.

Lisa's story may be an example of activity-based cognition (see below) and demonstrates the importance of meaning-in-activity for learning. It is an example of interdependence (see Chapter 16), because Lisa and the teacher worked together with tasks that were within Lisa's capacity for learning, in her zone of proximal development (see Chapter 2).

Simplifying goals

In this phase, it will often become necessary to simplify learning goals. This may be facilitated through optimization and compensation, rather than abandoning goals. For example, instead of teaching technical information about food nutrients, a student with JNCL may be taught which food sorts are healthy and which are not. Using personal experiences may support attention and memory, such as when learning how to make a weekly dinner menu or ensure a healthy diet for the family pet. When cognitive abilities decline, it is important to use topics of special interests and/or personal experience as foundations for learning. Evidence from other diagnostic groups suggests that dementia makes learning more difficult, but that learning is still possible (Clare & Jones, 2008; Clare et al., 2000; De Vreese et al., 2001; Kessels, Remmerswaal, & Wilson, 2011). However, it is likely to require frequent repetitions to refresh and maintain memories, more adaptation and more support.

The deterioration will not follow the same pace in different domains. In the present study (Appendix A), the mean age for onset of symptoms differed considerably between domains, for example between declines in vision, speech and gross motor functions (see Chapters 4, 6 and 7). Learning new skills may be possible in some areas but not in others.

Project work as educational strategy

In project work, students work together to produce a joint result and this result depends on students having different responsibilities. Project work makes it possible to utilize individual strengths and preferences, both when choosing themes for the project and when choosing tasks for the student with JNCL. This

strategy also ensures participation of the student with JNCL in a community of learners and being part of a social setting (Rogoff, Matusov, & White, 1996; Rogoff, Paradise, Arauz, Correa-Chávez, & Angelillo, 2003). Project activities may for example be applied to practical-aesthetic subjects such as woodwork and music. They may involve planning meetings where tasks and responsibilities are distributed among the participants, information gathering and making decisions about how to present the results. Several school subjects may be integrated into one project.

Ethel was interested in handicraft and had recently started to attend a handicraft workshop at the school together with her special education teacher. This was a useful preparation for adult life, because it was an interest that may be pursued well into adulthood. At the workshop, Ethel participated in making various items for decorating and practical use in the home. Some of the items were included in a sales exhibition, while other items would become Christmas gifts. Ethel's first task was to decorate Christmas cards to follow the gifts and be part of the sales exhibition. The teacher put small colored pieces of paper in front of Ethel, who crumbled them into paper balls, dipped the paper balls in glue and decorated the cards. Ethel still remembered colors and had conversations about colors with other people in the workshop. She wanted to use the best color combinations when she decorated the cards. The teacher helped Ethel find the colors she wanted. Another task was putting labels with product name and price on the products for the sales exhibition.

Changes in goal selection

Studies of the elderly population show that systematic educational effort may delay cognitive decline in dementia, given that the educational activities and tasks are manageable for this population (Baltes, 1997; Bird, 1998; Kawashima et al., 2015). Supporting maintenance is also an important aim of education for individuals with JNCL. There will gradually be less time spent on building new knowledge and skills, and more time spent on maintaining already established knowledge and skills. There are time windows for developmental progress and maintenance of skills. The role of the zone of proximal development, where education and support facilitate new learning and development, will gradually diminish. The zone of developmental maintenance however, where education and support contribute to maintaining knowledge and skills, will become more prominent (see Chapter 2 and 11). Appropriate daily cognitive stimulation is a part of the maintenance process. Goals can involve practical tasks, such as mixing ingredients when baking a cake, or more intellectual activities, such as discussing which cake ingredients are to be put

on the shopping list. Studies of elderly people with moderate dementia have found that participation in practical and cognitive activities lead to better performance of everyday tasks and increasing independence (Loewenstein, Acevedo, Czaja, & Duara, 2004). Similar strategies may be used with children and adolescents with JNCL, for example by stimulating and activating memories with familiar objects and sound recordings (Gylfason & Jóhannsdóttir, 2006; see Chapter 19).

Optimization and compensation

In this phase, children and adolescents with JNCL become gradually more dependent on help and support from other people and interdependence becomes more important in everyday tasks (see Chapter 16). However, participation seems to decrease when independence levels drop. Although help and support may be available most of the time, the survey found that social and societal participation becomes sparse (see Chapter 22). If activities are omitted from the weekly schedule, instead of being adapted to the person's abilities, this is likely to lead to a general reduction in activity level, less stimulation and a more passive life. For students with JNCL an important issue is to maintain activities and participation levels also when independence subsides. Optimization by forming and maintaining the means and activities to achieve individual goals should therefore be continued. It is further important to maintain the expectations of people in the environment even when extensive support is required. Education without expectations will be a degrading experience for most students, including students with JNCL (Elmerskog & Fosse, 2012).

Philip was 11 years old and his speech was starting to become slurred. One of the goals in Philip's Individual Education Plan was to maintain and strengthen his articulation skills as long as possible. A speech and language therapist visited the school once a week to give him training and give supervision to the teachers. The training program was followed-up by the teachers.

Because participation is an educational goal for students with JNCL, educational activities should be evaluated with regard to whether they support participation in activities that are manageable and meaningful for the student (Elmerskog & Fosse, 2012; Elmerskog & Storliløkken, 2006). Taking part in meaningful activities may have a considerable impact on the student's learning, cognitive maintenance and quality of life.

In this phase, the need for a clear daily and weekly structure will become evident (Elmerskog & Fosse, 2012). Structuring may be defined as a sequential organization of activities and tasks (see Chapter 16). A recognizable structure will

give the student an overview of the activities and may contribute to a feeling of security and control over the situation. If the student is able to read the daily schedule independently or interdependently, this may enhance feelings of self-efficacy and autonomy. Clear structures are also useful when adapting work situations or activities during leisure time (Elmerskog & Fosse, 2012).

Maintenance can be further enhanced through repetitions with some variety, and the students' personal knowledge and skills may be maintained through frequent refreshments of memory (Woods, Spector, Jones, Orrell, & Davies, 2009). The IEP should specify appropriate learning and maintenance goals, and how these can be achieved, for example how often repetitions and refreshments of memories of distant and recent events should be initiated by the teacher. Many individual education goals may be maintained through interdependence and participation (see Chapter 16). Over time, however, there will be skills that cannot be sustained, like writing in the example below.

Rachel was a young adult. She had been fascinated by fairytales and fantasy stories all her life. She had a rich imagination and a strong urge to communicate her thoughts. For some years she had been able to write her stories herself using a braille keyboard, but recently fine motor decline had made her writing pace slower and the number of spelling mistakes larger. Now her writing skills did no longer match her need and wish to express herself in writing. This activity of so great importance was maintained with compensatory measures. Rachel now conveyed her stories to the assistant who typed them on Rachel's computer. In this way, Rachel produced the stories and could listen to them via synthetic speech output, and she could share her stories with others.

The phase of severe dementia

Sustaining an active and stimulating life in order to achieve best possible life quality requires shifts in focus during the severe dementia phase of the disease. Skills will deteriorate in spite of efforts to maintain them, not abruptly but gradually over time. The zone of developmental maintenance replaces the zone of proximal development, and over time even the zone of developmental maintenance will shrink (see Chapter 2 and 11). Interdependence becomes clearer and participation in personally meaningful activities will gradually become the main source of communication and cognitive stimulation (see Chapter 16). When the time window for learning and maintenance begins to close, participation in meaningful activities may still be maintained through individual support from family and staff.

Hearing is less affected by the disease than the other sensory systems and the auditory system may function throughout life (Elmerskog & Fosse, 2012). The present study indicates that comprehension is better preserved than production of speech (see Chapter 6). The auditory sense and perception of speech are therefore important resources. In the phase of severe dementia, it may still be possible for persons with JNCL to perceive and understand environmental sounds and verbal information, such as descriptions of visual features, at least in relation to routines, everyday activities and other familiar events.

Optimization and compensation

Participation in activities with others will usually contribute to increasing learning and skill maintenance. Individuals with JNCL should therefore be in a rich speech environment to enhance and maintain their comprehension of the spoken language. They should be encouraged to listen and to speak when possible, as illustrated in the example below.

Richard was 18 years old and spent two days every week at a work place for persons with learning disabilities, together with a school assistant. Richard and the assistant worked interdependently: the assistant placed the material in front of Richard, and Richard's job was to disassemble a small piece of building equipment into two parts, while the assistant did the rest of the task and put the finished work away.

Around the work table was a group of 4–5 people, some of them very sociable and talkative. There was a continuous conversation and many themes were discussed. Richard had problems retrieving and articulating words, but he participated actively in the conversation by listening intently. Sometimes he would suddenly repeat a word or a name that someone had just said. Although it was usually very difficult to understand Richard's speech, he was understood because his utterances were relevant in the context and the words he said were repetitions of words that everyone had just heard. In this way, Richard participated in the conversation, both by listening and talking.

The presence of word-finding problems is a characteristic of severe dementia (see Chapter 5). In the story above, Richard was able to recognize words in the conversation and repeat some words now and then, but he was not able to bring new information into the dialogue. Asking open-ended questions to individuals with severe dementia is not likely to lead to relevant answers and may create stress and a feeling of shortcoming in individuals with JNCL. Instead, answering by choosing between a few possible answers depends more on word recognition,

which is easier than production (see Chapters 6 and 13). Students experiencing more difficulty can also be offered binary choices, a strategy that provides models of two possible answers, thus circumventing the need to retrieve words. Studies show that receptive vocabulary actually may continue to expand when other cognitive skills have started to decline (Adams, Kwon, Marshall, de Blicck, Pearce, & Mink, 2007).

For schools and other services, dealing with skill decline constitutes an unusual situation. There is a risk of a loss of participation opportunities provided to adolescents and adults with JNCL. For example, when their skills decline or they lose the ability to initiate actions for themselves, they might not be offered stimulating activities and situations. In addition, decline of skills may be misinterpreted as loss of interest and lead to non-participation.

Barbara was 20 years old and attended a work center for persons with learning disabilities. One of her favorite tasks was to make knitwear using a knitting machine. Barbara had a lot of strength in hands and arms and she was very proud of this. For a long time, she managed to control her movements perfectly when using the knitting machine. But eventually, problems occurred. Barbara was no longer able to regulate the strength needed to administer the knitting machine. Her arm movements became too abrupt and this destroyed the knitwear. The work center therefore decided that Barbara could no longer make knitwear on the knitting machine.

In the example above, the young woman lost a favorite work activity because of declining skills. Barbara and her assistant could have cooperated in doing the hand movements needed for knitting, and the favored activity and the associated social participation might have been maintained. Instead, compensational measures or interdependence were never considered. The next story illustrates compensation.

Brian was a young adult who had been interested in music all his life. When he was younger, he had been quite skilled at playing the piano. He gradually lost the motor control that was necessary for his playing. To compensate for this and help him to continue producing and performing his own music, he was given a computer program where he could compose music by choosing from a pool of free sounds and loops. With this program, Brian put together different instruments, sounds and rhythms together with helpers, and in this way he composed his own music.

The computer program enabled Brian to maintain his interest and ability in performing and producing music, even if he could no longer play the piano. His assistant gave the help and support that was necessary for keeping Brian engaged in music-making. Brian made all the creative choices needed to make the music. His music is shared and available for others on YouTube.

Ian and his assistant went by bus to an activity center five days every week. Each Tuesday Ian spent three hours in the kitchen together with a small group of other young people. The group collaborated in preparing food, and each group member had a different task. One of Ian's tasks was to administer the food processor. Some days Ian was more attentive and active than other days. On active days, he was able to find and push the on/off button himself, other days he needed to do this task together with his assistant. When they had turned on the food processor the assistant set a timer that would ring when it was time to turn the food processor off. On active days Ian shouted out when the clock rang to alert the others that the time was up.

Kitchen activities include a variety of tasks and sensory inputs. All group meals are social events, and when the cooking was finished the group would eat together around the table. For Ian, eating was restricted due to medical issues, but he could still have a little taste of the food. After the joint meal, Ian and his assistant brought food to other parts of the center. Most people will show enthusiasm when a nice young man offers them freshly cooked food, so there was a lot of positive feedback and enjoyable communication at this endpoint of the activity.

It is not clear if and when a person in this phase is no longer able to learn new skills or remember new information. Clinical experience suggests that learning is becoming restricted to areas of special interest and significant events in the individual's life. However, participation in diverse life situations may still be possible and is a prerequisite for learning and maintaining knowledge and skills, for creating interest and engagement, and for being part of the social and cultural community.

Mathematics education and visual impairment

Mathematical skills are used in many everyday situations. A practical understanding of mathematics enhances the individual's independence, and a basic understanding of mathematics is necessary for full inclusion in school and society. However, when experience is limited, mastery of mathematical skills

may depend on how these skills are learned, and they may not be mastered in all situations. One study found that 9–15-year-old street vendors in Brazil were able to solve mathematical problems when they sold fruit on the street, but not in relation to other activities, even if the problems by and large required the same level of mathematical skills (Nunes, Schlieman, & Carraher, 1993). Similarly, British children who have learned to work out pre-formulated math exercises may encounter difficulties when confronted with practical math problems in text form (Desforges, 1998). This indicates that mathematical reasoning may be grounded in concrete experiences, and at least in the early stages is activity-specific rather than context-independent knowledge (von Tetzchner, 2019). This may have implications for teaching and maintenance of mathematical skills.

Mathematics is an important school subject for all students, including students with severe visual impairment. Many students with blindness follow the grade curriculum when individual adaptations are sufficient and adequate. This however depends on teachers who have relevant knowledge, competence and access to necessary support (Rosenblum & Smith, 2012). Teaching mathematics requires mathematical and pedagogical competence (Ball, Hill, & Bass, 2005; Silverman & Thompson, 2008). Teaching mathematics to children with severe visual impairment depends on three elements that require additional competence. Firstly, teaching must be based on an analysis of mathematics as a discipline to ensure that it really is mathematics that is taught in class and not other school subjects. Secondly, severe visual impairment requires teaching methodology that compensates for the visual impairment, and the teacher must be familiar with this methodology. Finally, the teaching strategies must be adapted to the level of the individual student, to ensure an optimal curriculum (Ostad, 1982).

Children learning mathematics need to acquire some basic skills, including identifying differences and similarities, categorizing, ordering objects and quantities and understanding the principle of conservation (i.e., that a given amount or weight remains the same irrespective of the elements' size or shape). Basic skills also include the understanding and use of concepts that describe physical orientation of objects, such as «top,» «bottom,» and «beside» and concepts to compare quantities, such as «more», «less», and «the same.»

It is widely recognized that visual impairment highly influences learning of mathematics (Dick & Kubiak, 1997). Learning of basic mathematical concepts incidentally is largely based on the use of vision. Learning mathematical concepts without vision requires planned and formal teaching adapted to the individual's abilities and problems. «For children who are blind, direct teaching of mathematical concepts is essential; that is, the development of concepts must not be left to incidental learning» (Koenig & Holbrook, 2000, p. 374). However, many teachers find it difficult to teach mathematics to their students with visual

impairment, because much of the material used with the sighted peers is not useful. During the first school years, teaching mathematical concepts to children with typical development is mainly based on visual illustrations. Most of this material is difficult or impossible to adapt to tactile usage. To ensure that students with visual impairment have equal access to mathematics, teachers need at least a basic understanding of how mathematical concepts and operations may be presented and explained to students without sight. Coordination of teaching for both a child who is blind and the sighted classmates is important to ensure the child's inclusion in the class. This requires close cooperation between the main teachers and a special education teacher with specialization in visual impairment.

In spite of difficulties, many children with visual impairment do well in mathematics. One study of 248 students with visual impairment born between 1967 and 2001 found that 141 of the students (56.9 %) were taught mathematics on grade level. However, the study found considerable variation between students with different diagnoses: «Students with diagnoses related to the central nervous system had a comparatively higher risk of not attaining their normal grade level in mathematics» (Klingenberg et al., 2012, p. 93). Similarly, Erin and Koenig (1997) found that different causes of visual impairment lead to different spectra of symptoms, and that diagnoses with a neurological origin tended to affect both vision and learning in general.

Teaching mathematics to students with JNCL

Children with JNCL are born with normal vision and during the early years, they develop mathematical concepts in the same way as sighted children. Because of this, their understanding of mathematics concepts will be based on visual experiences when they start school. As their vision declines, they need gradually more adaptation to compensate for the loss of vision, although the conceptual foundation from the years with full vision gives them a good basis for further learning.

In the present survey (Appendix A), seventy-seven percent of the parents found that the mathematics education had a low impact on their child's daily life after school age. Mathematics had still been an important part of their curriculum. A small study of children and adolescents with JNCL found that most of them mastered all four arithmetical operations (i.e., addition, subtraction, multiplication and division) for a period of time, but only addition and the ability to count were still mastered in adulthood (Åberg, 2001). The present survey found that a few adults with JNCL were able to apply mental calculation in some situations, for example when playing cards or counting money. This indicates that mathematical

skills may be maintained within some activities, in line with the assumption that mathematical skills may be grounded and maintained in specific activities (von Tetzchner, 2019). One 25-year-old man with JNCL was reported to master all four arithmetical operations by using mental arithmetics. Another young man with JNCL was able to calculate with the help of a calculator. He had shown special interest in mathematics from early school years. This demonstrates that special interests may be utilized for teaching mathematics (Elmerskog & Fosse, 2012; see Chapter 16).

Students with JNCL differ when it comes to personal interests, strengths and abilities. Some of them have a persistent interest in mathematics. They show strong intrinsic motivation for exploring mathematical tasks and acquiring mathematical skills, which is a good precondition for learning. Knowledge about their challenges related to vision loss and about future decline should not reduce the teaching of mathematics to students with JNCL. On the other hand, as with children with typical development, there are children and adolescents with JNCL who are not interested in mathematics and find it boring or difficult. The content of the curriculum and the pace of teaching should be adapted to each student, so that their learning potential can be fully utilized.

Key skills

Children with JNCL have experiences and ideas about space and shape, and they have elementary knowledge about arrays and numbers. They have learned many words and concepts related to quantity and calculations. Children typically develop intermodal and amodal perception early in life (Bahrick, 2004) and get rich intermodal experience in construction play and other activities. They have counted objects and gradually understood that the last counted number is the number of objects in the set as a whole. Many students with JNCL have already started developing these key skills before the onset of visual decline. This gives them a good starting point compared to children who are born blind.

When children with JNCL start school, their understanding of the key mathematical concepts should be assessed before starting teaching mathematics. The tactile sense will become increasingly important when vision declines and students with JNCL should be given opportunities for learning to use the tactile sense to explore groups of objects with different characteristics with their hands. Access to physical manipulation of objects in the environment will enhance students' understanding of basic concepts (Csocsán, Klingenberg, Koskinen, & Sjöstedt, 2002). The tactile sense may be used by students with JNCL when counting and matching numbers to arrays of objects as well as finding matching quantities of objects that differ in size, shape and weight, although it may be

noted that also blindfolded sighted children have difficulties with non-visual recognition (von Tetzchner & Martinsen, 1980). The serial aspect is important for counting forwards and backwards, knowing the next number, matching one-to-one in counting, and counting in ones, fives and tens (Koenig & Holbrook, 2000).

During the first grades, sighted students make extensive use of pictures to enhance comprehension. Students who are blind must instead have access to tactile materials. For children with JNCL these key skills can be learned and supported by using tactile illustrations, bead strings or objects, or using vision for children with enough residual vision to locate and recognize the materials. Matching numbers to arrays of objects often requires extended learning opportunities in addition to adapted materials. Knowing number facts by heart will facilitate further learning. Number facts may for example be: $4 = 4+0$, $3+1$, $2+2$, $1+3$, and $4+0$. The multiplication table is another example. Automatization of number facts will help students to do calculations with more ease and efficiency, and the skills may be more resistant to future cognitive decline.

To adapt teaching to the needs of students with severe visual impairment, schools need support from visual impairment specialists, because ready-made materials are sparse. Mathematics is a complex domain and students with JNCL should learn basic mathematics skills early.

From she was little Sally liked numbers and mathematics became a favorite subject at school. During the first school years Sally learned a lot of number facts within addition, subtraction and multiplication before cognitive decline made further learning difficult. She continued to do calculations using this knowledge. Her mathematical skills declined in line with her cognitive decline. Addition was most resistant to decline. Eventually, she was no longer able to bridge ten but she could still do calculations like $12+5$. Doing calculations remained one of her favorite activities.

Students with JNCL will probably never be able to do advanced mathematics. Therefore, the basic mathematics curriculum for the first school years is the most important. Skills acquired early can continue to be part of the individual repertoire for many years, like for Sally above.

Linear algorithms for students with JNCL

An algorithm is a procedure and a specific way of writing mathematical problems. For sighted students, algorithms are often written using several lines to place the 1s in the 1-column, the 10s in the 10-column, and so on (see Figure 12.1). This is

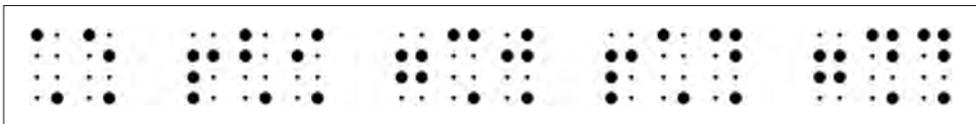
$$\begin{array}{r}
 12 \\
 + 27 \\
 \hline
 =
 \end{array}
 \qquad
 \begin{array}{r}
 43 \\
 + 17 \\
 \hline
 =
 \end{array}
 \qquad
 \begin{array}{r}
 31 \\
 + 42 \\
 \hline
 =
 \end{array}
 \qquad
 \begin{array}{r}
 57 \\
 + 22 \\
 \hline
 =
 \end{array}$$

Figure 12.1 From a visual workbook

functional for students who can see. When tasks are written in a book, students can get an immediate overview over the tasks on the page. Visual reading makes orientation on the book page very flexible.

For students with little or no vision, presentation of tasks like in Figure 12.1 will make orientation on the book page and doing calculations more difficult. For braille readers, tasks are most easily accessed by reading one line at a time. Algorithms should be linear, at least when teaching basic mathematics. Advanced braille users can learn several algorithms for doing arithmetic calculations, like the Nemeth Code (Nemeth, 1972) or the Unified English Braille (Cryer, Home, & Morley, 2013). Students with JNCL, however, might be better off continuing using the linear algorithms first learned.

Figure 12.2 shows one way a student with visual impairment might solve a task writing the calculation on one line in 8-dot braille (see Kacorri & Kouroupetroglou, 2013; see also Chapter 14). For children with JNCL with emergent cognitive decline the use of linear algorithms should be sustained.

Figure 12.2 $15+29 = 30+14 = 44$ in 8-dot braille

Mental math

«The ability to calculate mentally with efficiency is an essential skill for all students, but especially for students who are visually impaired» (Koenig & Holbrook, 2000, p. 388). Algorithms for sighted students include notation for every step in the calculation to support memory, like when bridging 10. Doing calculations without the use of vision is more taxing on working memory. Below are a few examples that show how students who are visually impaired may solve some mathematical tasks with the help of mental calculation. These strategies can of course be used by all students, not only students with visual impairment.

$33 - 19 = 34 - 20 = 14$ (adds 1 to each number to make the calculation easier)

$6 \times 18 = 60 + 48 = 108$ (multiplies the 10s and then the 1s)

$95 : 5 = 50/5 + 45/5 = 10 + 9 = 19$ (splits 95 to make the calculation easier)

For students with visual impairment, it is important to memorize the task while they do the calculations. If the student forgets the task, producing a repetition with synthetic speech or asking the teacher to say it again is more time consuming and less flexible than repeating by looking at the task like the sighted students do. When students read the task in braille, they can check the task independently. However, this requires orienting on the page or on the line and is not quite as easy as throwing a glance. Students with visual impairment thus have to use more of their working memory capacity while calculating. Students with JNCL develop problems with working memory but key skills and a basic understanding of arithmetic may enable them to do calculations within all four arithmetical operations (Åberg, 2001). Frequent repetitions may be required and a short mathematics session every day will probably facilitate learning and give better results than longer sessions once or twice a week.

Learning mathematic skills and maintaining already established skills may contribute to maintaining memory functions. There might be a double gain: memory supports learning of mathematics and learning of mathematics supports memory functions.

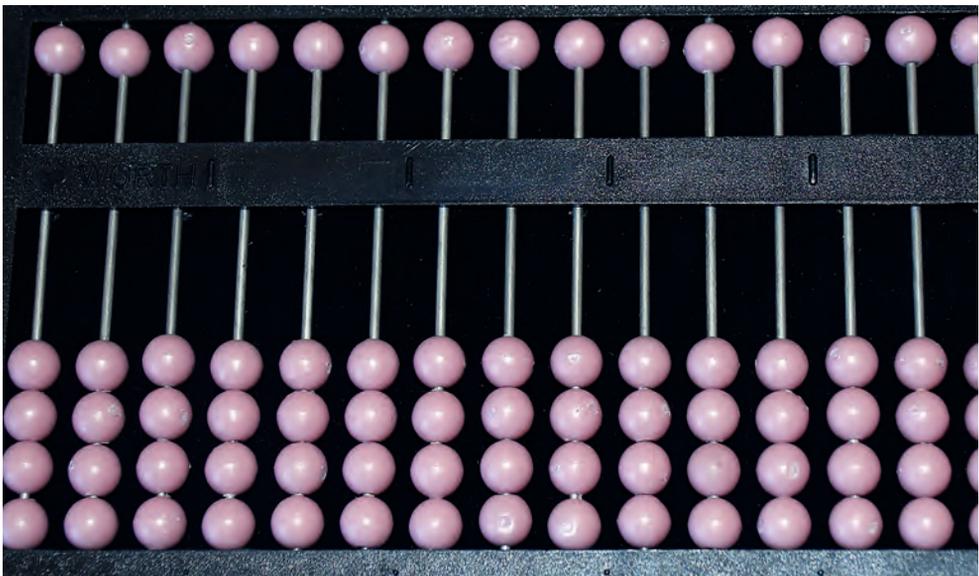


Figure 12.3 An abacus

Abacus and calculators

Abacus is a tool of Asian origin for doing arithmetic calculations which has been used in many countries for children with visual impairment, especially during the first school years. Abacus exists in many different designs (an example is shown in Figure 12.3). All four arithmetical operations can be done by using abacus. Using the abacus involves specific procedures which should be learned and remembered. The abacus should be introduced early to children with JNCL.

In the modern digital world, there are a number of assistive technologies which can be used in the education, including calculators and computer-based programs with braille or synthetic speech (see Chapter 19). Calculators with raised keys are usually easier to use for students with visual impairment than touch screens. Some calculators and computer programs can present the tasks in synthetic speech, the answers may be given orally with speech recognition, and the feedback is given with synthetic speech.

Practical use of basic mathematics

Teaching practical use of mathematics, like measuring time and money, is important for students with JNCL. These practical skills are connected with management of everyday situations and can easily be used to teach mathematics at different abstraction levels (Elmerskog & Fosse, 2012). Many everyday situations require use of mathematical skills, such as shopping, cooking, using a calendar with months and dates, following daily time schedules, measuring wood or other materials during art class, counting and finding specific amounts. Skills can be trained and afterwards repeated and maintained by being used in natural or adapted situations.

Every week the group makes a dish in the school kitchen. The group consists of eight students, but the recipes are written for four persons. The task is to solve the problem of rewriting the recipe from four to eight people and make a corresponding shopping list. And what happens when there are two extra joining the group for lunch?

Daily life contains many situations where basic mathematics skills are useful, such as when setting the lunch table for the appropriate number of persons at work place or at home, when filling the right amount of water and coffee into the coffee machine, or when playing Yahtzee (Yatzy) or other games, using dice with raised dots (see Chapter 21). When the learning capacity of individuals with JNCL is declining, they can still maintain and use basic mathematic skills that have

been learned and automatized. Assumptions about activity-specific mathematical knowledge also suggest that mathematics may be taught and maintained in specific activities.

Drama in the education of students with JNCL

«By engaging in experiences within drama and the expressive arts, children and young people will recognize and represent feelings and emotions, both their own and those of others. Drama plays a central role in shaping our sense of our personal, social and cultural identity» (Scottish Government, 2009). This aim of the Curriculum for Excellence (Scottish Government, 2009), to recognize and represent feelings, has particular significance for persons with disabilities as they try to make sense of their place in their environment and maintain connections with others.

With the onset of visual impairment, young people with JNCL are likely to become less aware of eye contact, as well as facial expressions, body posture and gesture. Young people with JNCL will need support to maintain their understanding of non-verbal forms of communication, and drama may play an important role in this process. Drama may offer opportunities for the young persons to use their spoken language and vocalizations to explore the meaning of words, sounds and expressions whilst interacting with others. However, as the condition progresses, they will have increasing difficulty imagining themselves in someone else's environment and it can be a real challenge for them to imagine themselves in a different body and in a different context. They may also find it increasingly difficult to accept that an adult is going into a role and sometimes have difficulty with the difference between reality and pretense. As such it is important that the teacher is sensitive to the young persons' needs and challenges them appropriately. If young persons are struggling to perceive their own world, it may be very confusing to try to eject them from that and imagine themselves in the newly created world within the drama lesson. It is important that young people with JNCL feel safe within the drama structures. A second approach lies in the introduction of pupils to the experience of literature, not simply as a vehicle for teaching communication but to provide access to a "cultural heritage" (Grove & Park 1996; Grove 2012).

Experience from working with young people with JNCL indicates that involvement in drama relies on getting the young people to engage with their emotions, with their heart. If they get completely caught up in their learning, they will be more likely to remember the learning and maintain these skills in

communication. Young people with JNCL might require material and tools in other modalities to get this engagement, but with the right support they can participate and benefit from drama throughout their education.

The first step: Finding a good story.

At the Royal Blind School, student engagement in drama has often involved adaptations of many of the classics, including works by Shakespeare (Macbeth, Hamlet, The Tempest), Charles Dickens (Oliver Twist, David Copperfield, Great Expectations, Ebenezer Scrooge), Mark Twain (Prince and the Pauper), Robert Louis Stevenson (Treasure Island), and many others.

Classics are classics because they are great stories with timeless themes that pull the actors and the public in on an emotional level. They can be delivered on the same level of a child's story like «The three little pigs». Why do the three little pigs with a young adult when it is possible to do «Macbeth» or «Great Expectations»? There is no reason that a young person who has JNCL should be doing fairy stories for younger children (although there is nothing wrong with fairy stories in the right context). The key is to find ways to make appropriate material accessible, engaging, fun and educational. And the classics can be adapted for a whole range of needs and abilities (Grove, 2012; Grove & Park, 1996).

For example, «Macbeth» is a story that starts with two friends, Macbeth and King Duncan. There is a war and Macbeth is loyal to Duncan. Duncan rewards him with a new castle and a ceremony. All is well and the relationship has been set – perhaps over three or four drama sessions. Then Lady Macbeth decides they should do something nasty to King Duncan, something that Duncan does not deserve. This has real emotional impact because the story is built on the friendship of Macbeth and Duncan; the students care about them and they are appalled at the injustice. It really gets the students going, they get motivated to speak out and be involved in the drama! And then when Macbeth doesn't get caught but the guards do! The students jump up and down – they are hooked on the story. They can learn all sorts of things. About relationships, about expressing themselves, about emotions, about things not going the way they should. But being involved in the drama, in pretend, means there is a distance, it is not their real life. They can be engaged in a deeply emotional way, but it is not necessarily their emotion. When the drama closes, they come back to themselves and they can discuss how they felt.

Of course, in real life, they will never be involved in a brutal murder with daggers – that is the joy of the story. But they will engage in those related emotions. They can experiment with them and play with them. This is valuable and especially for young persons with JNCL who may be increasingly struggling to manage their own emotions.

Some strategies used in drama class

Hot seat

First of all, a good introduction to drama is the use of the «hot seat». The hot seat is a chair or an area, and once you sit there, you become a character and need to stay in character. This way, students learn about physically going into character and have a very clear cue to start the drama. It is important to remember that for young people with JNCL, there is no eye contact or gesture for beginning the drama as one would with sighted students. This physical change of position works.

The hot seat area can be adapted to any situation and the use of additional props can help the young person get into character. For example, if the person in the hot seat was supposed to be a giant, a ladder could be set up in the hot seat area to give the impression of height and the voice would come from above! Another hot seat might be covered in fur to make it the king's throne or they might just sit on the floor with an old, tattered, smelly coat if they were supposed to be a sad, old homeless woman. The important thing is the physical movement from one area to another, in effect, stepping physically into role and touching the material objects that are related to the role. Remember that young people with JNCL will not be able to recognize standard cues for readiness or acceptance of role play by eye contact, body language or gestures. The physical change to the hot seat acts as the cue instead.

Teacher in role

The teacher can go into role as a character and interact with the class. This way, the teacher can change the direction of the drama, challenge thinking and manage the group within the drama. This keeps true to the drama, without having to be a «teacher» giving instructions.

And depending on the class, the teacher in role can really explore issues and emotions in a grittier way. As long as the students are secure in the convention, they can really enjoy having an encounter with Bill Sykes (from «Oliver Twist») who is threatening, violent and has a foul temper, even if this could induce tears in other circumstances! They can experiment with emotions and empathy and explore how characters react to different situations. Real life issues like bullying or being isolated in society can also be addressed. They can test this out in a completely safe environment.

This leads on to improvisation. Through the «Teacher in Role» exercise, the young people will have been given ideas about appropriate vocabulary and ideas and this can help the young persons have the confidence to take forward the improvisation and use it themselves.

In the context of a story, young people with JNCL can be supported to experiment with what it is like to yell and shout and stamp the feet and identify when these emotions are appropriate and how people react. They can explore what it is like to be overcome with anger. What does it feel like to be threatened, frightened, in a situation where something is not right, for example Bill Sykes? How does it feel when someone comes close into your face, invades your space and speaks in a threatening manner? This is pretend, it is fun, it also touches the emotions and it is hugely valuable learning. This can act as a major stress release for a student – especially if they do not really understand their symptoms and cannot articulate how they are feeling.

Freeze frame and thought tap

Drama can focus on emotions, understanding of own and the emotions of others. What do emotions feel like? How do they look? How do other people perceive you? This may help students with JNCL to understand and make connections. Again, the classics are a great tool for exploring these themes. For example, in working on «The Tempest,» a «freeze frame» of facial emotions was created to explore the different characters. Using the cue of a tap on their shoulder, the young person has to speak or vocalize in character. If it is an angry freeze, for example they might say, with conviction «*I hate him!*» or just an angry noise. This may connect the group, give them ideas and reassure them that they are all doing the same work.

In this connection, scripts are sections that are good to work with. In «Macbeth» the script for the Witches scene is particularly engaging: «*When shall we three meet again in thunder lightning or in rain...*» It does not seem to matter whether the students understand the intricacies of the language. The words are poetic and flow, and it is nice to say them. This scene also allows one to build a great atmosphere in the room, using sound effects, music, voice, candlelight as the witches are telling Macbeth he will be King.

Young people with JNCL do not need to have a physical script if this is too much, they can work from a CD or use the support of an assistant prompting lines quietly.

Using materials

The use of tactile material and physical objects within the drama structure is very important to make it a more sensory experience. Connection with a physical symbolic object helps the young person with JNCL relate to a character (e.g., the use of a fur covered chair for a king's throne). It is important to use real objects and props that have meaning and are authentic. Why use a plastic apple when a real one feels and smells so totally different? «Real» props link to real experiences.

A creative approach to physical movement can also build bridges for communication within the drama. A small working area in some scenarios can give the young person a real experience of being restricted and having no freedom. For example, they can really crawl into a cramped corner to denote a cave or be under thick blankets for an air raid shelter. A total freedom of movement in other situations like being outside in a grassy ditch for a scene on a battlefield gives a very different experience and generates other responses. The use of space is hugely important to help develop and maintain the young persons' spatial awareness and these experiences help them want to keep exploring.

It is also useful to record voices or film in class. This can re-create and remind the young person of the actual physical experiences in which they participated. In a way it is similar to using sound like a photograph to open up a rich set of memories associated with being in that drama. This can be important for them, and also can be a means of assessment and self-evaluation for the young person.

Conclusions

Exploring real and imaginary situations may help young people with JNCL to understand and share their world. If they can get emotionally involved and care about the outcome, they may be more creative and strive to be actively involved in the story, independently or interdependently (see Chapter 16). But the advantage of drama is that the structures enable there to be enough distance to allow the students to be engaged in the feeling without being swept up with the emotion. They can empathize with the characters without getting upset themselves. That is the power of drama!

References

- Åberg, L. (2001). Juvenile neuronal ceroid lipofuscinosis: Brain-related symptoms and their treatment. Doctoral dissertation, University of Helsinki, Faculty of Medicine.
- Adams, H. R., Kwon, J., Marshall, F. J., de Blicke, E. A., Pearce, D. A., & Mink, J. W. (2007). Neuropsychological symptoms of juvenile-onset batten disease: experiences from 2 studies. *Journal of Child Neurology*, 22, 621–627.
- Baines, S., P. Saxby, P., & Ehlert, K. (1987). Reality orientation and reminiscence therapy: A controlled cross-over study of elderly confused people. *British Journal of Psychiatry*, 151, 222–231.
- Ball, D. L., Hill, H. C., & Bass, H. (2005). Knowing mathematics for teaching: Who knows mathematics well enough to teach third grade, and how can we decide? *American Educator*, 29, 14–22, 43–46.
- Baltes, P. B. (1997). On the incomplete architecture of human ontogeny: Selection, optimization, and compensation as foundation of developmental theory. *American Psychologist*, 52, 366–380.

- Baltes, P. B., & Baltes, M. M. (1990). Psychological perspectives on successful aging: The model of selective optimization with compensation. In P. B. Baltes & M. M. Baltes (Eds.), *Successful aging: Perspectives from the behavioral sciences* (pp. 1–34). New York: Cambridge University Press.
- Bahrick, L. E. (2004). The development of perception in a multimodal environment. In G. Bremner & A. Slater (Eds.), *Theories of infant development* (pp. 90–120). Malden, MA: Blackwell.
- Bills, W., Johnston, L. W., Wilhelm, R., & Graham, L. (2011). *Teach and be taught: A guide to teaching students with Batten disease*. Reynoldsburg, Ohio: Batten Disease Support and Research Association. Retrieved June 1st 2012 from <http://www.bdsra.org/programsservices/bdsra-publications/on>
- Bird, M. (1998). Clinical use of preserved learning capacity in dementia. *Australasian Journal on Ageing*, 17, 161–166.
- Clare, L., & Jones, R. S. P. (2008). Errorless learning in the rehabilitation of memory impairment: A critical review. *Neuropsychology Review*, 18, 1–23.
- Clare, L., Wilson, B. A., Carter, G., Breen, K., Gosses, A., & Hodges, J. R. (2000). Intervening with everyday memory problems in dementia of Alzheimer type: An errorless learning approach. *Journal of Clinical and Experimental Neuropsychology*, 22, 132–146.
- Cryer, H., Home, S., & Morley, S. (2013). Unified English braille in the United Kingdom: Part I – Examination by technical expert braille users. *The British Journal of Visual Impairment*, 31, 228–237.
- Csocsán, E., Klingenberg, O., Koskinen, K.-L., & Sjöstedt, S. (2002). Maths «seen» with other eyes. A blind child in the classroom: Teacher's guide into mathematics. Esbo: Schildts Förlags.
- Damon, W., & Phelps, E. (1989) Strategic uses of peer learning in children's education. In T. J. Berndt & G.-W. Ladd (Eds.), *Peer relationships in child development* (pp. 135–157). New York: John Wiley.
- Desforges, C. (1998). Learning and teaching: Current views and perspectives. In D. Shorrocks-Taylor (Ed.), *Directions in educational psychology* (pp. 5–18). London: Whurr/Wiley.
- De Vreese, L. P., Neri, M., Fioravanti, M., Belloi, L., & Zanetti, O. (2001). Memory rehabilitation in Alzheimer's disease: A review of progress. *International Journal of Geriatric Psychiatry*, 16, 794–809.
- Dick, T. & Kubiak, E. (1997). Issues and aids for teaching mathematics to the blind. *Mathematics Teacher*, 90, 344–349.
- Elmerskog, B., & Storliløkken, M. (2006). Beyond the classroom. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 31–35). Örebro, Sweden: Swedish Institute for Special Needs Education.
- Elmerskog, B., & Fosse, P. (Eds.) (2012). *Lære for livet: en innføring i pedagogikk og læring ved juvenil neuronal ceroid lipofuscinose (JNCL)* (Learning for life. An introduction to education and learning in juvenile neuronal ceroid lipofuscinosis). Trondheim: Akademika forlag.
- Elmerskog, B., Storliløkken, M., & Tellevik, J. M. (2008). Fra helhetlige perspektiver på habilitering og rehabilitering til funksjonelle og individrettede tiltak (From comprehensive perspectives on habilitation and rehabilitation to functional and person-oriented interventions). In P. Fosse & O. Klingenberg (Eds.), *Pedagogiske og psykologiske perspektiver på opplæring av synshemmede* (pp. 87–95). Melhus: Snøfugl Forlag.
- Erin, J. N., & Koenig, A. J., (1997). The student with a visual disability and a learning disability. *Journal of Learning Disabilities*, 30, 309–320.

- Fasting, R. B. (2013). Adapted education: The Norwegian pathway to inclusive and efficient education. *International Journal of Inclusive Education*, 17, 263–276.
- Grove, N. (Ed.). (2012). *Using storytelling to support children and adults with special needs: Transforming lives through telling tales*. London: Routledge.
- Grove, N., & Park, K. (1996). *Odyssey now*. London: Jessica Kingsley.
- Gylfason, K. B., & Jóhannsdóttir, K. G. (2006). Living with Batten disease in Iceland: The story of Hrafnhildur. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 48–49). Örebro, Sweden: Swedish Institute for Special Needs Education.
- Kacorri, H., & Kouroupetroglou, G. (2013). Design and developing methodology for 8-dot braille code systems. In C. Stephanidis & M. Antona (Eds.), *Universal access in human-computer interaction: Applications and services for quality of life. Proceedings of International Conference on Universal Access in Human-Computer Interaction, July 2013* (pp. 331–340). Berlin: Springer.
- Kawashima, R., Hiller, D. L., Sereda, S. L., Antonczak, M., Serger, K., Gannon, D., et al. (2015). SAIDO learning as a cognitive intervention for dementia care: A preliminary study. *Journal of the American Medical Directors Association*, 16, 56–62.
- Kessels, R. P. C., Remmerswaal, M., & Wilson, B. A. (2011). Assessment of nondeclarative learning in severe Alzheimer dementia: The Implicit Memory Test (IMT). *Alzheimer Disease and Associated Disorders*, 25, 179–183.
- Koenig, A. J., & Holbrook, M. C. (Eds.) (2000). *Foundations of education: Instructional strategies for teaching children and youths with visual impairments, Volume II*. New York: AFB Press.
- Loewenstein, D. A., Acevedo, A., Czaja, S. J., & Duara, R. (2004). Cognitive rehabilitation of mildly impaired Alzheimer disease patients on cholinesterase inhibitors. *American Journal of Geriatric Psychiatry*, 12, 395–402.
- Nemeth, A. (1972). *The Nemeth Braille Code for mathematics and science notation*. Louisville, KY: American Printing House for the Blind.
- Norris, C., & Closs, A. (1999). Child and parent relationships with teachers in schools responsible for the education of children with serious medical conditions. *British Journal of Special Education*, 26, 29–33.
- Nunes, T., Schlieman, A. D., & Carraher, D. W. (1993). *Street mathematics and school mathematics*. Cambridge, UK: Cambridge University Press.
- Ostad, S. A. (1982). *Lærerveiledning til matematikk når barnet ikke ser: Begynneropplæringen* (Teacher guide to mathematics when the child does not see: Beginning education). Oslo: Tiden.
- Rogoff, B., Dahl, A., & Callanan, M. (2018). The importance of understanding children's lived experience. *Developmental Review*, 50, 5–15.
- Rogoff, B., Matusov, E., & White, C. (1996). Models of teaching and learning: Participation in a community of learners. In D. Olson & N. Torrance (Eds.), *The handbook of education and human development* (pp. 388–414). London: Blackwell.
- Rogoff, B., Paradise, R., Arauz, R. M., Correa-Chávez, M., & Angelillo, C. (2003). Firsthand learning through intent participation. *Annual Review of Psychology*, 54, 175–203.
- Rosenblum, L. P., & Smith, D. (2012). Instruction in specialized braille codes, abacus, and tactile graphics at universities in the United States and Canada. *Journal of Visual Impairment and Blindness*, 106, 339–350.
- Schoenberg, M. R., & Scott, J. G. (2011). Cognitive decline in childhood or young adulthood. In M. R. Schoenberg & J. G. Scott (Eds.), *The little black book of neuropsychology: A syndrome-based approach* (pp. 839–861). New York, NY: Springer.

- Scottish Government (2009). Curriculum for excellence: Expressive Arts – Principles and practice. Downloaded from: <https://education.gov.scot/Documents/expressive-arts-pp.pdf>
- Silverman, J., & Thompson, P. W. (2008). Toward a framework for the development of mathematical knowledge for teaching. *Journal of Mathematics Teacher Education*, 11, 499–511.
- Tellevik, J. M. (2008). Kognitive, sosiale og emosjonelle konsekvenser av synstap (Cognitive, social and emotional consequences of vision loss). In P. Fosse & O. Klingenberg (Eds.) *Pedagogiske og psykologiske perspektiver på opplæring av synshemmede* (pp. 71–85). Melhus: Snøfugl forlag.
- Uvebrant, P. (2006). The interaction between neurology and education in juvenile Batten disease. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 95–97). Örebro, Sweden: Swedish Institute for Special Needs Education.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- von Tetzchner, S., Fosse, P., & Elmerskog, B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta 1832 (2013)*, 1894–1905.
- von Tetzchner, S., & Martinsen, H. (1980). A psycholinguistic study of the language of the blind: I. Verbalism. *International Journal of Psycholinguistics*, 7, 49–61.
- Williams, L. A. (2008). Students first: A trans-disciplinary team approach to the education of a student with Batten Disease. Retrieved from Internet August 17th 2018. https://ir.canterbury.ac.nz/bitstream/handle/10092/2647/Thesis_fulltext.pdf;sequence=1
- Woods, B., Spector, A. E., Jones, C. A., Orrell, M., & S. P. Davies, S. P. (2009). Reminiscence therapy for dementia (Review). *The Cochrane Library*, 2009, No. 1.

Communication and Language in Education and Intervention for Children and Adults with Juvenile Neuronal Ceroid Lipofuscinosis

Anne-Grethe Tøssebro, Stephen von Tetzchner, Anna Christal, Deirdre McElrone, Mary Siggs, Judith D. Oxley and June Cameron

Young people with juvenile neuronal ceroid lipofuscinosis (JNCL) will gradually experience a change from speaking effortlessly and fluently to finding speaking to be tiresome and even frustrating (see Chapter 6). It is not possible to stop or prevent the disease from developing. Speech problems eventually will have a major impact on the young person's social life and life situation in general. The promotion of communication and language is therefore always an important aim in the curriculum of students with JNCL. Interventions may contribute to strengthen and maintain language skills, prolong the period with fluent and intelligible speech, and compensate for loss of spoken language. There is a need for developing strategies to prepare for and compensate for the speech decline but there are few studies on language and communication intervention for this group.

The present chapter discusses interventions for maintaining intelligible speech as long as possible and the use of augmentative and alternative communication (AAC) to supplement and compensate for the impairment of speech.

Interventions to strengthen and maintain intelligible speech

The aim of speech and language therapy is to strengthen the child's speech and language skills, and may include preventive measures, diagnosis, education, training, and counseling. Specialist services with speech and language therapy exist in all the countries participating in the project. Interventions related to fluency, and to strengthening and maintaining the functions of breath, voice and articulation, are relevant for students with JNCL, as are interventions related to communication strategies.

Interventions will not prevent symptoms from developing but clinical experience indicates that decline can, to some extent, be postponed. Gayton (1982) used rhythm strategies to improve speech fluency and intelligibility in two students with JNCL, such as accompanying the speech with clapping the hands or talking in time with a metronome. However, the cognitive problems made it difficult for the students to follow the strategies and they were therefore abandoned. Experiences from Norway suggest that strategies used in stuttering therapy might lead to temporary improvement in fluency and maintenance of speech intelligibility (von Tetzchner, 1993). Structured and systematic training may establish a foundation to meet the upcoming symptoms, and speech and language therapists may give useful advice to parents and schools about training strategies.

In the present survey (see Appendix A) parents were asked about speech and language therapy. Forty-three parents answered that their child had received speech and language therapy (see Figure 13.1), and that speech therapy was first provided at an average age of 9.9 years (SD 5.6). The survey did not ask for details about the speech and language therapy.

The parents were also asked to evaluate the effect of the therapy on a scale from 1 (no impact) to 5 (very high impact). The average evaluation score was 3.2 (SD 1.2, range 1–5). Eighteen respondents (41.9%) answered that the therapy had a high or very high impact, and 11 respondents (25.6%) answered that the effect was moderate. Fourteen parents (32.6%) found little or no effect of the speech and language therapy (Figure 13.2). These findings suggest that a considerable part of the respondents found speech and language therapy useful for their child, and

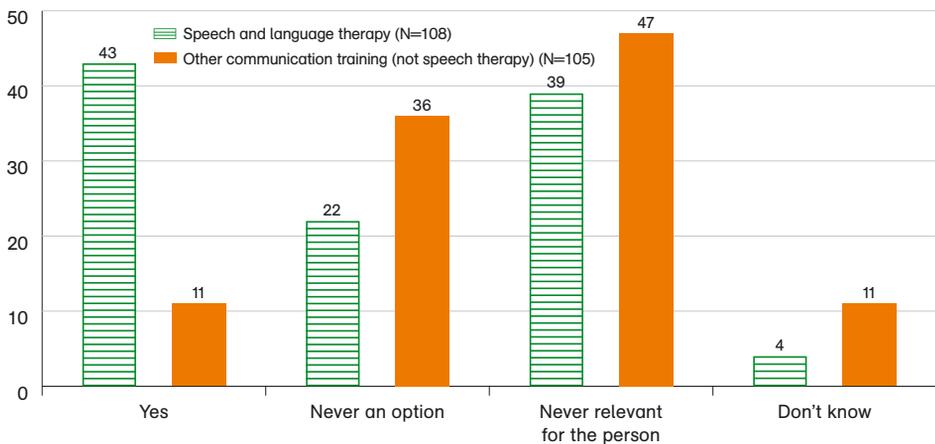


Figure 13.1 Number of individuals with JNCL who had received speech and language therapy or other communication training

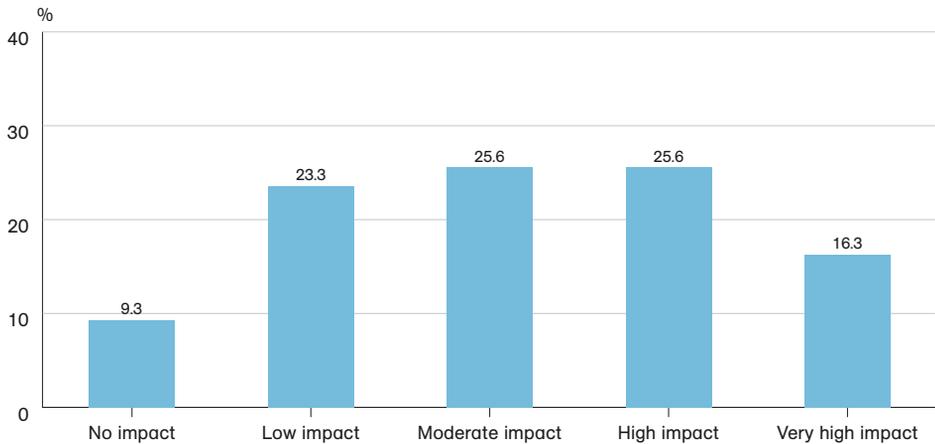


Figure 13.2 Impact of speech and language therapy (percent of answers) (N=43)

that speech and language therapy for some children and young people with JNCL may be useful for a shorter or longer period.

Only 11 participants had received communication training other than speech and language therapy (see Figure 13.1), and this was provided for the first time at an average age of 12 years (SD 6.1). Five respondents answered that the training had a high or very high impact, and two that the impact was moderate, while four found little or no impact of the training. The number was small and the evaluation varied but seemed to indicate that communication training other than speech and language therapy may be useful for some children and young people with JNCL.

Augmentative and alternative communication (AAC)

Augmentative and alternative communication (AAC) is an umbrella term that includes diverse communication modes other than speech for individuals with impairments of language and communication, including manual signs, graphic symbols and tactile communication systems. During the last decades, AAC has become widely used for children and adults with severe speech or language disorders. AAC can be an alternative or a supplement to speech. The function of AAC is to support communication and language skills in general and should preferably be available throughout the day and in all situations. Today, children and adults with different diagnoses use AAC (von Tetzchner & Martinsen, 2000).

Unaided and aided communication

AAC can be divided into two broad groups: unaided and aided communication. Unaided forms of communication are *produced* by the user. There is no need for any device but unaided forms of communication require some degree of motor ability. They may include vocalizations, facial expressions, gestures, manual sign systems and sign language. Communication partners must be able to interpret and understand the communicative expressions, and the message is often produced as a collaboration between the AAC user and the communication partner (Clarke & Wilkinson, 2009; Hörmeyer & Renner, 2013; Solomon-Rice & Soto, 2011).

Aided forms of communication are *selected* by the user. They imply some kind of device, such as a folder, a board, a book, or an electronic device with synthetic speech output, usually with graphic symbols or letters and words (von Tetzchner & Martinsen, 2000). Aided communication thus depends on the availability of a communication aid, and availability may be limited in many situations (von Tetzchner et al., 2018). Because most communication aids contain pictures and graphic symbols, they usually require some degree of vision but it is possible to adapt them to a tactile recognition or auditory scanning. For children with JNCL, the selection of expressions should be as simple as possible, so the expressions can be used for communication also in later phases of the disease with dementia.

Objects may function as *tangible* or *tactile* symbols (and visual for children with residual vision), often called "objects of reference" (objects used in a symbolic manner to refer to something). It is important that it is not the function of the object itself that determines how it is used but the category it represents, of people, animals, things, activities, and so on (McLarty, 1997; Park, 1995, 1997). For some they are a step on the way towards more advanced symbol learning and literacy. Objects of reference may be an alternative to graphic symbols. Braille can also be used in aided communication (Park, 1997; Rowland & Schweigert, 2000).

Both aided and unaided communication can function as alternatives to speech or to augment speech. Families and professionals need to work closely together when choosing an AAC mode. If possible, an interdisciplinary team should be involved in the decision-making process, as well as in planning, implementation and evaluation of the intervention. The communication form or forms that are chosen should be based on an assessment of the individual's strengths and weaknesses. To make the environment communicatively accessible, the communication partners, too, need training (von Tetzchner & Stadskleiv, 2016). If a child or young person with JNCL learns manual signs because speech is slurred and difficult to understand, the family, staff and peers must be taught the same sign vocabulary. Different forms of AAC can be combined, both with each other and of course with speech.

User groups

Various forms of AAC have been developed for persons with limited spoken language and are used with children and adults with a variety of diagnoses, in particular with motor disorders, intellectual disabilities, severe language disorders and autism spectrum disorders. Users may have age-appropriate comprehension and problems with expression only, or problems with both receptive and expressive language. Some children with severe and multiple disabilities need AAC to develop any communication skills. There are three main groups of AAC users (von Tetzchner & Martinsen, 2000).

- 1) The expressive group is characterized by a significant gap between comprehension and production of spoken language. Many in this group have motor impairments but also children with Down syndrome may belong to this group.
- 2) The language support group may need AAC temporarily to support the development of spoken language, or as a lifelong supplement to speech.
- 3) The alternative language group includes children and adults with little or no comprehension and production of spoken language. AAC becomes the main communication form for both them and their communication partners.

People who use AAC differ considerably in language comprehension and this has implications for the intervention. «For persons with good comprehension, developing conversation skills is the most important educational goal. Lack of language skills is not the cause of their limitations (...), but lack of capability to use what they understand» (von Tetzchner & Martinsen, 2000, p. 277).

When AAC is part of the intervention, communication partners must know the person's repertoire of communication expressions and strategies. AAC may affect communication fluency by slowing down the communication pace, and it is important to give the child or adult enough time to participate in the dialogue. Manual signs, objects of reference or other aided expressions may be used in unusual and idiosyncratic ways (see also Chapter 6). In an inclusive environment, it is essential that all potential communication partners, adults and peers, become familiar with the special elements in conversations involving AAC. Otherwise, the AAC user may become restricted to communicating only with a few persons, mainly adults. Measures should thus be taken to ensure communication with all relevant persons in the environment, including peers.

AAC and JNCL

In early childhood, children with JNCL have typical language development but as the disease progresses, many gradually become difficult to understand even for those who know them well. In the later phases of the disease, comprehension also becomes more affected by the cognitive problems (see Chapter 6). The main reason for offering AAC to children and young people with JNCL is their increasing gap between comprehension and production, even if comprehension also tends to decline.

AAC for children with severe visual impairment must be adapted to compensate for the vision impairment. AAC may include tactile symbols, gestures, manual signs, object of reference, communication boards and books, and electronic devices. Most communication aids are designed for sighted children or adults, and have graphic symbols (e.g., PCS, Pictograms, Blissymbols), photographs or drawings, but it is possible to equip electronic communication aids with tactile symbols or a braille keyboard and synthetic speech (Goldware & Silver, 1998; Rowland & Schweigert, 2000). Information about AAC for children who are visually impaired is available at resource centers or special schools for students with visual impairment. In Norway, *Tactile symbols and plans* provide information about tactile adaptations of communication aids (Grini & Aasen, 2013). *Tangible symbol systems primer* (Rowland & Schweigert, undated) is available for free download.

A literature search revealed almost no descriptions of the AAC for children or adults with JNCL, the case history below is an exception.

A case history

A 17-year-old girl was provided with an electronic communication aid with speech output when her speech was beginning to become unintelligible (von Tetzchner, 1992). When the communication aid was introduced, she had good comprehension of spoken language and a large vocabulary. Her speech was intelligible to her family, friends and teachers, and many of her utterances were comprehensible for people who did not know her as well. At that time, she only occasionally experienced communication breakdowns and misunderstandings, but because the ability to learn was declining, it was necessary to start then, before she really needed the aid. The overall intervention goal was to help the girl maintain social interaction and conversations for as long as possible. This goal was the basis of all intervention strategies.

Hello	I want something	Hungry	I	Person	Help	Finished	Goodbye
I want to say something	Try to guess	Thirsty	Before	After	Music	Radio	When
I must talk this way	More	Tired	In	Out	Read	Weave	Stupid
Yes	Almost yes	Again	Wrong	Don't know	Wait	Almost no	No

Figure 13.3 The girl's first vocabulary

Aid and vocabulary

Because the girl was blind, she was provided with an electronic aid with speech output. When she pressed a square on the overlay, the device produced the word. At that time, Norwegian synthetic speech was not well developed, so digitized speech was used. The vocabulary was recorded using the speech of a young girl of the same age and using the same dialect. In Norway, technical aids are provided free by the State, and the communication aid was borrowed from the local technical aid center. The girl had a large vocabulary, which she would not be able to express through use of an aid with a limited vocabulary, because she was unable to learn navigation to select a large number of lexical items. Therefore, the intervention strategy was not to give her a means for expressing all her needs, interests and ideas, but to use the communication aid to guide the conversational partner to infer what she wanted to say. Some of the words were chosen to gain attention, some to define conversational domains, and some to direct the inferences of the partner. To help her navigate, some of the words were marked tactually on the communication aid.

The initial vocabulary is shown in Figure 13.3. Because of her good language skills, it was believed that the girl could learn 32 words and phrases, and possibly more later. However, even a vocabulary of 32 items proved too difficult. It was difficult for her to find what she wanted to say and in collaboration with the girl, the vocabulary was reduced to nine items by omitting words and combining topics into one utterance.

The selection of vocabulary was an unhurried process where she participated actively together with parents and teachers. She was a member of the project team, and it was an important motivation for her that the project may be of help to others with the same disease as herself.

Use of the aid

When the girl got the communication aid, she did not need it to make herself understood in conversations, but it still had a positive impact on both her communication skills and her life situation. She immediately took a liking to it, named it "Talking Lady", and demonstrated it willingly to family members, friends and helpers. She had great expectations with regard to what it could do for her.

The girl experienced some communication difficulties before she got Talking Lady. Sometimes she was misunderstood or not understood at all, and the conversation broke down. However, conversation failure was too sensitive for her to discuss and she refused to talk about it. Talking Lady represented a means for her to manage her difficulties and made it possible for her to talk about her problems, and to discuss how to cope with them. This influenced the interactions positively and improved the communication without the girl actually using the device for communication. The inability to talk about the communication problems had been a significant stress on the family, and by catalyzing openness and communication, the device led to a better family situation. According to her family, the positive effects had already given more benefits than the efforts invested.

With time the girl's speech became more difficult to understand. Sometimes even her mother was not sure whether she said yes or no. Such situations were resolved by the mother asking more yes-no questions, and the Talking Lady was never really used for communication. But it remained a psychological support.

AAC use among children and adults with JNCL

In addition to speech and visual problems, support of expressive language needs to take into account decline in motor and cognitive functioning. Different symptoms become noticeable at different ages, and there is considerable individual variation (see Figure 1.1 in Chapter 1). The fact that speech problems often become noticeable before other motor problems may imply that communicating by using hands and arms might be maintained longer than speech.

When the parents in the present project were asked about AAC or special communication strategies, eleven parents mentioned that they and the child used some kind of alternative means in communication. One family mentioned that their daughter with JNCL used to squeeze their hand to say "yes" and "no", another child clapped the hands to express herself, although it was not always clear to the family what this meant. A third young man would nod and smile or frown to indicate "yes" and "no". A young woman coughed when she needed to go to the toilet. Some families used assisted auditory scanning, they listed several items and waited for a yes or no. A few families had tried electronic devices with synthetic speech output, but the results were rarely positive.

They tried some communication programs on the iPad, but they were all visual and she was blind, so they were fun for the teachers but not useful for my daughter.

However, both awareness and use of AAC and support with alternative means seem to be increasing. When the parents were asked about AAC intervention, twelve parents mentioned using specific gestures or manual signs, six mentioned objects of reference, eight mentioned electronic devices (including switch-operated devices), and eleven parents mentioned "other strategies". Most of the participants were still in the process of learning, and for some the training had been abandoned. Some mentioned that AAC might be introduced as a precautionary measure. One parent commented:

Alternative communication methods should be provided for youths with JNCL (and children) earlier (age 10–15), as they still have a good opportunity to learn and adapt to new things.

In summary, the results of the present survey indicate that most young people with JNCL develop severe problems with expressive speech. Only a small number of children, adolescents and adults with JNCL have been given opportunity to learn alternative communication strategies, but such strategies seemed to become more common in this group. There is also a greater focus on trying out these strategies, like the manual sign project below. AAC training can be provided by speech and language therapists or by other professionals.

Manual signs

Manual signs and gestures may be useful when the ability to speak declines. Manual signs are rarely used in interventions for people who are blind, partly because of their inability to learn through visual observation. However, teaching methods can be adapted to compensate for the missing visual sense, for example using signs in the tactile modality (Downing & Eichinger, 1990). Manual signs may be taught to students with JNCL (and the other students in their class), with the aim of establishing a communicative competence that may be useful later in life if speech becomes unintelligible.

Manual signing as AAC does not mean a national sign language with full sentences and correct grammar, but rather signs from a manual sign system like Signing Exact English, Signalong, Irish Láhm or Signed Norwegian, representing familiar objects, persons, activities or events. This usage of signs is called

key-word signing or sign supported speech (Budyanto, Sheehy, & Khofidotur, 2018; Glacken et al., 2018; Grove & Woll, 2017). In many countries, there are websites with sign dictionaries, which include videos of signs. These websites are useful for parents and teachers when they are choosing signs. In addition, there are many apps available for mobile phones or tablets that can provide the sign for a word immediately (in various sign languages or manual sign systems). Most people who provide support for individuals with JNCL are not familiar with manual signs. Having an app handy can help them see how the sign is produced (via a video) so they can then instruct the child. Pictures of signs in books can be hard to follow, and books are not always available.

As the disease progresses, language ability gradually declines, expressive speech more than comprehension (see Chapter 6). The communication partners will speak when communicating with the young person with JNCL. They do not need to use signs while speaking, because the child with JNCL will not be able to see the signs. However, successful communication will depend on their understanding of the child's signs. Signs may not only be learned in special sessions – the partners may show the student with JNCL new signs or help him or her articulate known or new signs in natural situations.

Alternative methods with signing on the body include *Tactile Signing for Sensory Learners* (Tassels) (Woodall & Charnock, 2017) or *Canaan Barrie on-body signs*, developed by Mary Lee and Lindi MacVillians at the Royal Blind School in Edinburgh (von Eichwald, 2015). Manual signs articulated on the body may be easier to learn for some students, because they are designed for people with a visual impairment and additional learning needs.

Manual signs may be easier to articulate than speech. The individual uses the hands and body and does not depend upon the availability of a communication aid. As motor problems in hands and arms appear relatively late in most persons with JNCL (see Chapter 7), manual signs could prolong the period of successful communication. Manual signs may have the same flexibility and diversity as spoken languages. It is possible to create idiosyncratic "home signs" when needed and the signs can be adapted to the motor skills of the person (Grove, 1990; Rudd, Grove & Pring, 2007). Signs can both augment and substitute for spoken language and can be combined with communication aids or other forms of AAC.

During recent years, projects on manual signs have been conducted in the Nordic countries for a small number of children with JNCL. The following is a short description of a Norwegian project with four participants with JNCL.

A project on manual signs

The aim of the project was to investigate if students with JNCL could learn manual signs and use them spontaneously for communication. The project was organized by Statped (the national Norwegian agency for special education support) and took place from September to December 2014. The project started with a two-day course for families and staff to enable the schools and families to start teaching manual signs to the students. The course included information about communication problems for individuals with JNCL, use of AAC and how to teach signs to students who are not able to imitate by using vision, and where to find sign dictionaries on the Internet. The families and staff chose a sign repertoire for each student and decided what signs should be taught first.

Methods

Four students, aged 8, 14, 17 and 19 years, participated in the project (the names below are not their real names). Information about sign use was collected with the help of three registration forms that had been reviewed during the course, and a short interview with staff and/or families. The forms were completed by parents and/or staff daily. Educational counselors from Statped visited the participants during the project period. They were also available for families and staff on email and telephone during the three-month project period.

Results

At the closing time of the project, information from the registration and interviews indicated that the students had learned to perform a varied number of signs and could produce the signs when asked to do so. This was a step in the learning process towards communication. The students had also used signs on their own initiative a few times to ask for or talk about something. This suggests an emergent understanding of signing as communication. One student, Arne, initiated communication by using signs several times. During a visit from the resource center, another student, Christer, spontaneously signed DRINK to tell that he wanted something to drink.

Table 13.1 shows that the results varied a lot between the children. Arne was the youngest and learned a substantial number of manual signs during the project period. He and his family attended a one-week seminar, where they received guidance from a resource center for sign language. This was a major support for Arne and his family, and the center gave much more input and assistance than the other families and staff had access to.

Since the project period ended, Arne has continued learning signs and is signing quite a lot. He signs fluently as part of his communication along with speech. He finds it helpful, especially after he experienced episodes with severe

Table 13.1 Manual signs taught to the participants in the Norwegian sign project

Arne 8 years	VOLCANO, STAR WARS, SPACESHIP, GALAXY, PLANET, PIRATE, SWORD, PISTOL, LIGHT SABER, WALKIE TALKIE, HANDCUFFS, POLICE, THIEF, PRISON, SPY, ZOO, TORCH, REFLECTIVE VEST, BIKE HELMET, LEGO, DUPLO, HEADPHONES, AUDIO BOOK, DAISY PLAYER, RAT, SVEIN (NAME), SEWAGE, JAMES BOND, SKI, PLAY CARDS, PLAY, RAIN, SNOW, ICE, LAVA, MONDAY, TUESDAY, WEDNESDAY, THURSDAY, FRIDAY, SATURDAY, SUNDAY, PLATE, KNIFE, FORK, SPOON, BREAD, BUTTER, SUGAR, LIVER PASTE, JAM, GRAPES, ORANGE, APPLE, BANANA, CUCUMBER, HAM, TEA, COFFEE, EGG, MILK, COCO, CHOCOLATE, JUICE, SODA, STRAWBERRY JAM, BLUEBERRY JAM, RASPBERRY JAM, CHEESE, BROWN CHEESE, WHITE CHEESE, RED, ORANGE, WHITE, YELLOW, BLUE, PURPLE, PINK, GREEN, BLACK, BROWN, DARK BROWN ETC, LIGHT BROWN ETC, HOUSE, STONE, SUN, TREE, MOUNTAIN, HEAVEN, RIVER, TUNNEL, GRAFFITI, WINDOW, DOOR, HEART, TOMBSTONE, CHURCH, HELLO, GOODBYE, THANKS FOR FOOD, HUNGRY, THIRSTY, GLASS, CUP, DRINK, TEASPOON, GOOD NIGHT, GOOD MORNING, BOOK, WATER JUG, COFFEE POT, CAT, DOG, TIGER, LION, SNAKE, COW, RIDE, SHEEP, GOAT, TROUSERS, SWEATER, JACKET, SHOES, SOCKS, HAT, FEATHER, GLASSES, TOGETHER WITH SISTER/BROTHER, MAYBE, TODAY, YESTERDAY (PAST), TOMORROW (FUTURE), MOTHER, FATHER, CHILD, TEACHER, TO TEACH, TO LEARN, FLOUR, ICING SUGAR, GLAZE, CAKE, MELT, RUBBER SPATULA, PLASTIC, SHEET, WINDOW, ONE, TWO, THREE, FOUR, FIVE, SIX, SEVEN, EIGHT, NINE, TEN, ELEVEN – TWENTY, HUNDRED, THOUSAND, MILLION, YATZY, CLOWN, EYE, NOSE, MOUTH, TOMATO, SALT, KETCHUP, THE WHOLE ALPHABET, GARDERMOEN, BERGEN, TROMSØ, OSLO, FILM, CD.
Bente 14 years	HELLO, GOOD MORNING, YES, NO, MUM, DAD, MUSIC, GOODBYE, PAIN, COFFEE, HOME, SUN, SIBLING, TEACHER.
Christer 17 years	FOOD, DRINK, YES, NO
David 19 years	YES, NO, QUIET, WAIT, WHAT, STOP.

stuttering that made communication difficult for him. Another student stopped learning signs shortly after the project period ended, because of declines in memory and fine motor skills. One student had learned a few signs before the project started and expanded his sign repertoire with three signs during the project period. For one of the students there is no information about signing after the project ended.

Factors related to signing

The reasoning behind introducing manual signs for children and young people with JNCL was the possibility that signing might reduce the communication problems related to stuttering, articulation problems and general speech decline (see Chapter 6). Articulating manual signs is less complex than articulating words,

and this may contribute to maintaining motor skills related to signing longer than the motor movements of speech. This was demonstrated by Arne.

It is conceivable that signing may reduce the word finding problems typical of dementia, but it is not obvious that it is easier to recall a manual sign than a spoken word. In addition, memory for newly learned manual signs might not be as robust as memory for spoken words acquired much earlier in development. The parents observed that Arne used signs when he had difficulties remembering the words. This might indicate that manual signs somehow is easier to recall or that he has two ways of expressing something and can choose the other way when the first does not work. Having alternative modes may thus facilitate communication through cross-modal stimulation (von Tetzchner & Martinsen, 2000). These are important issues to investigate further.

Factors of importance for AAC intervention

Many factors may determine whether AAC intervention will lead to positive learning results, including administrative and educational practices, and political and ideological attitudes and regulations.

Daily learning and practicing opportunities

Learning depends on practice, and AAC should be used during the whole day. Learning will be delayed if AAC is only used with the teacher who is responsible for the training. After intervention has started, the use of AAC should be encouraged in different situations and with different communication partners, within and outside educational settings. Some forms of AAC, for example manual signing or using a technical communication device, will require some formal teaching. A brief formal teaching session should be scheduled for every day, preferably early in the teaching program. Such daily repetition may be necessary to ensure progress and maintenance. Communication partners should have the training necessary to be able to give help and guidance when needed. Opportunities for communication should always be emphasized and periods when acquired communication skills cannot be understood should be prevented.

To ensure that AAC becomes a natural and functional part of the child's communication, the child should take part in a variety of situations where signing or other forms of AAC can be used in a natural way. Such situations can be at school with teachers and peers, or at home with family. Preferably, the young person with JNCL should be able to use AAC in all common situations. Because young people with JNCL often use short utterances (speech and AAC) in everyday

situations and conversations, communication partners must be able to recognize the person's repertoire of manual signs or other AAC modes, know how these are used by the person, and infer their potential meaning in the context. This applies to family members, staff and peers.

Barriers in the learning environment

The pathological processes involved in JNCL gradually reduce the ability to learn. However, these processes are not the only factors that may influence learning negatively. There can also be barriers in the learning environment. Schools might be reluctant to implement new educational strategies, especially when these strategies deviate from the usual guidelines of the curriculum, such as teaching manual signs to children who are blind. Schools can also focus too much on their own lack of competence. AAC may be seen as a difficult domain for teachers if the school has no prior experience with AAC. This can create resistance from staff and prevent the search for possibilities and competence. Sometimes there are disagreements concerning responsibility, whether an intervention is a matter for the educational or health authorities, or a specialized service outside the school (for a thorough discussion, see Goldbart & Marshall, 2004).

Other barriers concern structures and systems, for example related to resource allocation, time needed for staff to attend courses or get external guidance. Lack of communication structures for information transfer within the school or institution and rigid staff schedules can be barriers. In one of the schools participating in the Norwegian manual sign project, only one assistant was involved in the intervention. The assistant was very competent, but her schedule only allowed signing sessions twice a week. This was probably too little for an optimal learning process.

It is important to identify and overcome barriers in the learning environment. Teaching AAC is not really very complex or time consuming. Solutions should be sought to secure the student's learning and development as fast as possible. Students with JNCL should not have to wait for clarifications while the disease continues progressing. It is usually possible to find solutions if there is willingness to change and discussions are unrestricted and creative. Support from specialist services can be an important factor to help getting started.

Learning for the future

For young children with JNCL, the need for AAC will usually lie many years ahead. At the same time, it is important to start teaching early in order to utilize the child's best learning capacity. Resource centers are sometimes asked about the

motivation behind AAC intervention when the child or adolescent still speaks well. Enhanced and precautionary learning are basic principles when preparing AAC intervention for students with JNCL (see Chapter 12). All students engage in learning for the future. The knowledge and skills acquired by children and adolescents constitute a foundation for a successful life in society. To ensure learning, the goals and teaching methods applied are adapted to the students' age and capacities. Students are not usually aware of why they should learn the specific skills and knowledge presented in the classroom, but they learn anyway. Learning for the future is a basis for all students.

Inclusion

The principle of inclusion is important in all the countries participating in the present project. It is an overall aim included in the legislation of many countries that education should take place in a community of peers. Learning in a social setting may in itself be a strong motivator (see Chapter 12). At the same time, students have different needs and abilities. The learning environment must be adapted to give students learning opportunities, irrespective of their abilities and challenges. This implies that the objective and methods will vary among students with special needs, but ideally, this should not compromise their social affiliation.

Communication is important for participation, and declining communication skills can make inclusion difficult. This is the motivation for implementing measures that may strengthen the communication skills of students with JNCL. These measures are likely to make demands on both the child with JNCL, his or her peers and the school. When a student with JNCL is learning AAC, the class or group also need to learn AAC to maintain his or her participation and communication with the peer community. This is illustrated in the story below.

A young boy with JNCL, "John", was communicating with a combination of speech and manual signs. His signing was fluent and functional, and it was obvious that signing was very beneficial for his communication. It was however, not always easy for communication partners to understand what John wanted to say. He attended a school where all the students in his group had learned key-word signing. The boy's teachers had a saying that illustrated their view on the learning environment of his group: "A life necessity for John is also good for the other students and completely harmless for everybody".

The teachers' saying is a good and simple guideline for inclusion.

Selecting AAC vocabulary

When choosing AAC vocabulary the individual's preferences, interests and needs must be in focus. The words that are selected should be meaningful and useful for him. Individual preferences include words for special interests and favorites, activities he often wants to do, and activities he often wants to talk about.

Talking about emotional events is important. The vocabulary should enable the person to communicate about current and earlier events, as well as other important life events, and express emotions like happiness, pride, grief and fear. For some individuals, emotional events can be preferred and recurrent topics of conversation. Vocabulary should also include activities that happen routinely, daily or weekly. The person may for example want to ask whether he is going to do a particular activity, when it will happen or who will be joining him.

Play and games

For children in primary school, play and games often give extra motivation for activities and schoolwork. A board game developed in Norway, «Eltho Tactile», consists of a board, dice and activity cards. The teacher decides what tasks should be asked for on the activity cards, for example «Show the sign for Star Wars», «Sign the names of your siblings» or «Show the sign for your favorite activity». The tasks can be written in braille if the student has the necessary reading skills, or in print for the staff to read aloud. The potential for individual adaptations is considerable.

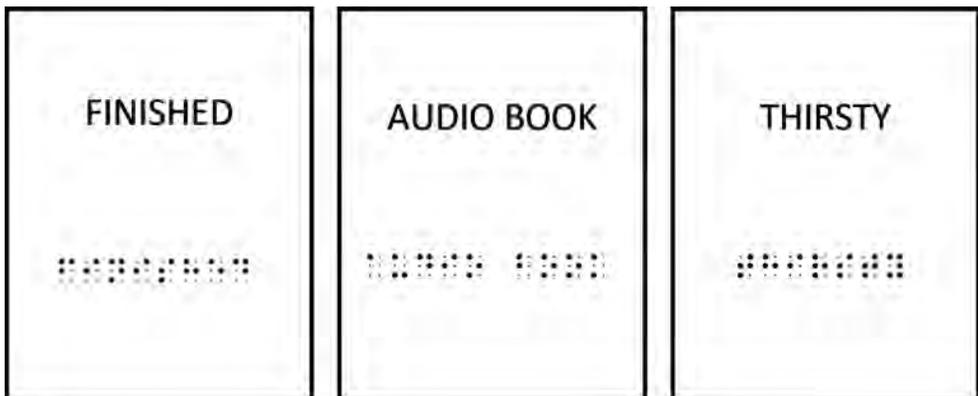


Figure 13.4 Cards with braille and print used for teaching manual signs

In the Norwegian sign project, Arne's family developed several kinds of material for teaching and maintaining signs. The parents made cards with words written in braille and print (see Figure 13.4). The cards had holes and were strung on a rope, and the rope was hanging across the room. When Arne followed the rope, every now and then there would be a card showing a word and his task was to sign the corresponding sign. The parents would count the number of correct signs: «You remembered 16 signs out of 20!» or it could also be turned into a competition: «You remembered 16 signs and I only remembered 14!» Cards that were mastered were marked with a tactile sticker to help both Arne and the family to keep track of the signs he had learned.

These kinds of activities can be used together with others, for example family, friends, teachers or classmates (see also play and games in Chapter 21).

AAC and transitions

Transitions often represent challenges for inclusion and participation. Children and young people with JNCL may attend a new class and meet new classmates. Staff also changes. Young adults go through the transition from being a student to attending a sheltered workshop or a day center (see Chapter 23). For many there is also a transition from living with the parents to moving into a residential home with services and support from staff. In adulthood, speech is often severely affected by the disease and difficult to understand for peers and staff who are not familiar with the person's communication and history. When preparing a major transition, the individual's needs and wishes should be the foundation of the planning process. New communication partners will need information about the person's history, interest and preferences, and about present communication skills, including AAC. There is a considerable risk that an individual with JNCL may be underestimated if the new staff are not aware of the gap between the person's comprehension and production of speech, and his sometimes idiosyncratic way of using words and alternative expressions.

Some useful tools to promote successful communication

As communication becomes more difficult for the individual with JNCL, several tools may be useful. Some of the tools have technical equivalents, for example computer programs or electronic communication devices (see Chapter 19).

Partner-assisted auditory scanning

This technique utilizes comprehension in supporting expressive language and may be useful in supporting communication in persons who have severe motor and speech impairments (Clarke & Price, 2012). The technique supports memory by presenting some alternative responses when the person with JNCL has difficulties finding the words or the expressive means are very limited. It may be useful for persons with severe visual impairment because it relies on the auditory sense.

In partner-assisted scanning, the communication partner presents a sequence of utterances, one of which the person might want to say (see procedure in Table 13.2). The utterances might be «I want to talk about something», «I want something to eat», «I want something to drink», «I want to do something», or «I want to tell you how I feel». All the categories are presented and then repeated one by one with pauses to wait for a possible response from the person. When the person has chosen a category, a sequence of alternatives within the category may be presented in the same way – first the whole sequence and then the alternatives one by one with pauses. The number of utterances presented can be adapted to the cognitive level of the person.

Comments from several parents in the present project indicated that they had used partner-assisted scanning with their child when speech was becoming difficult to understand, and some had developed homemade systems based on the same principle.

Table 13.2 Routine for partner-assisted scanning

• Ensure that the person is well-positioned and comfortable.
• Get the student's attention by saying his/her name.
• Tell the student what you are going to do: "I'm going to give you (3, 4 or 5) choices. First listen carefully to all the choices".
• List the choices slowly and clearly, for example: "music....book....video....something else".
• Make sure you still have the person's attention.
• Say: "[Name], tell me when you hear the one you want".
• List the choices in the same order as before, pausing slightly between each of them.
• Watch and wait for the person to give a response.
• Repeat the person's response, for example: "You said _____".
• If the response is unclear, tell the person you are having difficulty reading the response and repeat the sequence again.

I decided that the best strategy was auditory scanning, where I would say individual items in a list and wait for her to respond yes or no.

When it became difficult for him to explain/remember what he wanted to say, we parents developed a 'Question system' to define the subject and afterwards continue questioning about that specific subject.

I was responsible for training her in auditory scanning.

We use a question technique to find out what she wants to talk about, because otherwise we cannot understand what she says.

For persons with JNCL, the only prerequisite for using partner-assisted scanning is the ability to express "yes" and "no". This can be spoken words or vocalizations, gestures, signs or body posture. If it has become difficult for the person to indicate "yes" and "no" the communication partners need to be observant and sensitive. It may be a very subtle movement indicating a positive or negative response, as described in these examples.

He would nod his head or give us a big smile when we gave him the desired words or requests or he would frown or tighten his brow if we were off the mark. Later we would give him only two choices at a time so he could use "yes" or "no" to make his desired choices.

Pressing the hand once: "yes", pressing the hand twice: "no".

To be sure all communication partners know how a person expresses "yes" and "no" when the expressions are atypical or idiosyncratic, it is practical to have a photo or written description of these expressions. Partners may also benefit from pre-made category sets made by professionals, that can guide strategic use of the «20 questions» technique. Partners sometimes panic in the moment, if the child needs to communicate urgently, and give the child random options. When given a predetermined set of categories to present to the child, systematically, partners can be more confident and helpful playing their role. The child with JNCL benefits from the presentation of categories that are relevant, and not randomly determined by the partner in the urgency of the moment. Additional memory support is provided through the use of the fixed order because it benefits expectation and recognition.



Figure 13.5 Objects of reference used on an electronic device with speech output

Objects of reference and time support

Objects of reference can be used to support choice making, memory, structuring of the day, orientation and communication (McLarty, 1997; Park, 1995, 1997). It is important that the objects that can be attributed a symbolic function are introduced at an early stage, so that the young person becomes familiar with using them. The objects can for example represent people, animals, objects, locations, activities or events. They can be whole objects, parts of objects or miniatures. Objects of reference can also be attached to electronic communication devices with speech output (Figure 13.5), enabling the person to express something, make comments and social greetings even if he is unable to speak with his own voice.

It is vital that the person can understand what is happening during the day and, when possible, make choices about what to do and when. In the early stages of the disease, the person may be able to use timetables in braille or Moon. Linking objects of reference to the text on the timetable may pave the way for using them when the person can no longer read braille or Moon (see Chapter 14).

When persons with JNCL reach the stage when they find it difficult to remember the timetable of a full day, the time table can be simplified by giving cards with a tactile symbol or an object of reference for only the upcoming and the next activity, for example for physical education while saying «Now physical education» and the symbol or object for lunch while saying «then lunch». If

considered useful there can be a "finished box" to put tactile symbol cards and objects for activities that have been finished. There is considerable literature on this strategy and it can be accessed readily online by using the key words such as *calendar box* or *anticipation box*.

Chat book

Chat books enable the person to remember what he has done recently, and to communicate about these events and activities. They are also known as *remnant books* (Marvin & Privratsky, 1999), or *conversation books*. Whenever the person does something memorable or experiences something interesting or amusing, it can be made into a story. The story can be written in a book with a souvenir or memento attached, enabling the person to choose which story he wants to talk about (see Figure 13.6). In this way, the person may remember events and activities and share information about things that have happened. The chat book needs to be updated regularly. The person knows that his communication partner has read the story and therefore will understand the conversation, even if the person's speech is not easily understood. A parent described the use of a chat book.

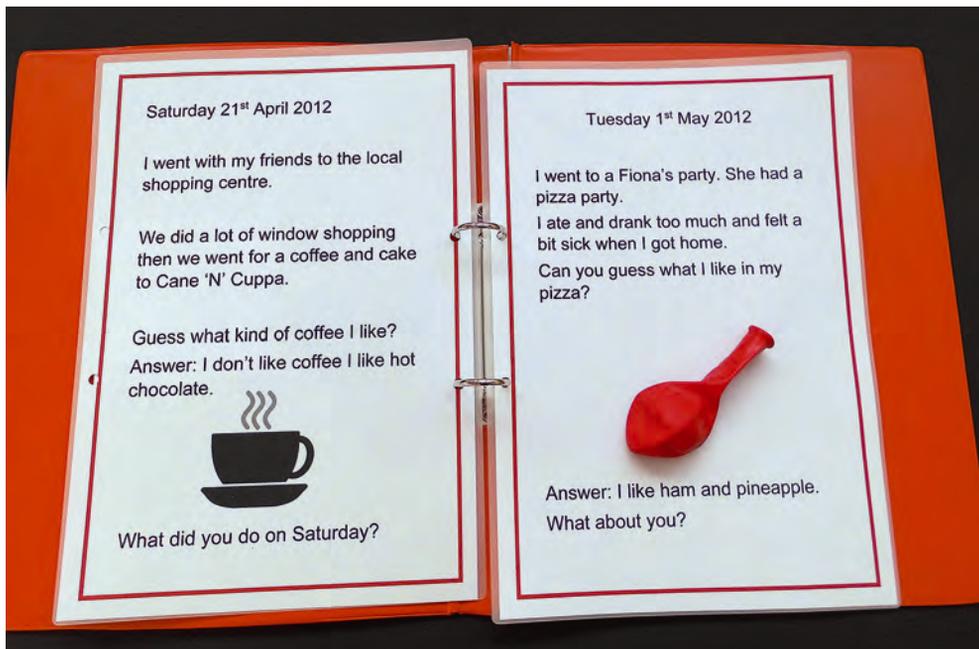


Figure 13.6 A chat book

Key ring with laminated sheets that describe something about her. For example five red sheets about school, five white sheets about the family. Every sheet gives some information about her and by using the ring, you can ask her further questions about the subjects.

At a later stage, the book may be supplemented with audio recordings, which the person can listen to, or even be recorded on to a computer or an electronic communication device, enabling him to share the stories independently.

Memory/experience book

A memory book is another way to enable the person to activate memories and communicate with people about things that they have done and which are interesting and meaningful to them. A memory book can contain stories about the person's life and include recordings made over time of activities, friends, music, and so forth (Figure 13.7). This is a more permanent and long-term record of the person's life. The book should be created jointly by everyone who knows the person well, particularly family and friends.

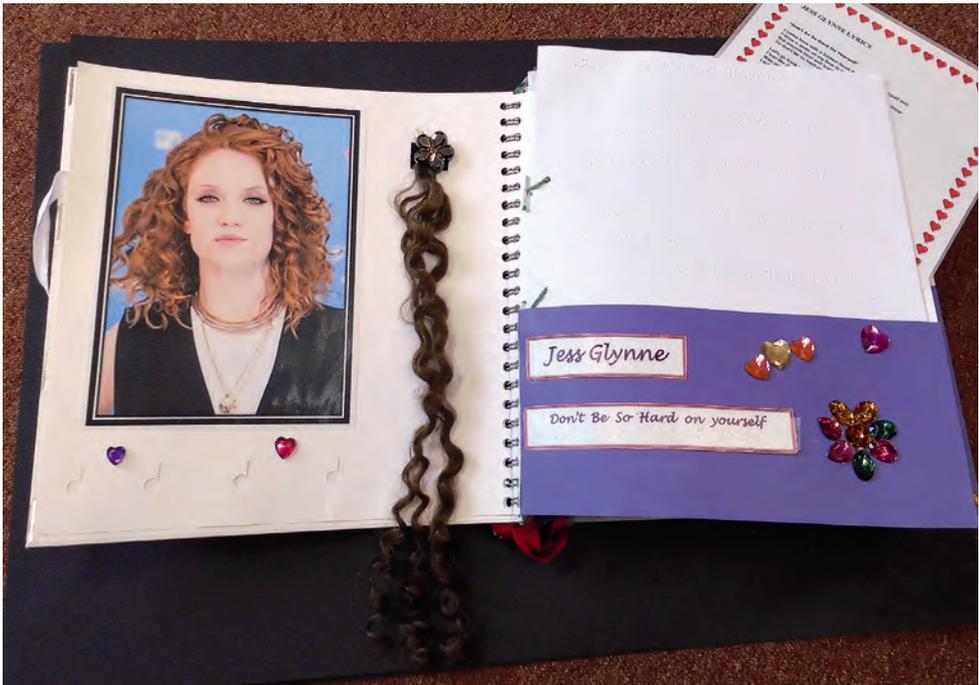


Figure 13.7 Music page of a memory book

Communication passport

The communication passport is usually a small booklet with the most important information about the person (Millar, 1998). This may be a useful tool for people who are not familiar with the person, for example hospital staff or support staff, to quickly learn some relevant biographical information, like the things that the person is interested in and the most effective ways to communicate with him. It describes how the person shows emotional states and physical needs, for example sad, happy, tired, hungry, and thirsty or need to go to the toilet. It is written in a simple, easily accessible style in the first person and will help people to understand and relate to the person rather than his problems or disability.

The passport needs to be updated regularly, so that it contains current, relevant information about the person's needs, abilities and interests. Photos can be included to illustrate how to support the person or to provide communication partners with topics of conversation.

Community request cards

Community request cards are a simple way of helping persons communicate more effectively in the wider community with people who are not familiar with their communication support needs (Figure 13.8). People supporting the person use all available communication methods to find out what he wants to say in a particular situation, for example what he wants to order in a café or make an appointment at the hairdressers. The support person then writes a short note outlining this information which the person with JNCL gives to the person he wants to communicate with. This enables members of the public to know exactly what the person wants even if they do not understand his speech or other forms of communication. It makes the person the center of the interaction, rather than the support worker or family member, and the communication partner is more likely to address the person with JNCL directly in return.



Figure 13.8 Community request cards

Prioritizing communication

The findings from the present project show the importance of giving priority to communication and starting early. It is not possible to predict the individual course of the disease, but young people with JNCL will develop problems with expressive speech sooner or later. AAC should therefore be considered. The learning capacity is best early in life, before speech problems start developing. Teaching AAC therefore should start early and preferably before speech starts to decline or has become severely impaired. At the same time, there are large individual differences and some young people with JNCL probably will be able to learn AAC in adolescence or even adulthood, as illustrated in these parents' story about their son:

We began asking for evaluation of a communication device when he was 12 on the advice of a Batten researcher. Unfortunately, the school did not get on the ball and get anything done until he was around age 16. He is 18 now and really just starting to use the device.

Some staff members also mentioned that they had tried alternative means of communication:

When speech and facial expressions were lost, we tried to invent dialogues hoping to create the ideas, which she wanted. That was only possible because we knew her well. It was nearly impossible to invent new signs etc., because she could not keep them in mind. Only when she knew them from before we could use them.

I have always thought it was a pity for her having been deprived of different aids and initiatives – because many educational measures were offered when it was too late to be of any use for her – because it gets more and more difficult to learn new things as the disease progresses.

The need for future research

Both the findings from the present project and clinical experience indicate that augmenting communication for individuals with JNCL can be important for sustaining their life quality. The ultimate goal is to give persons with JNCL some extra years of fluent communication when possible. However, there is a need for more systematic knowledge. The present JNCL project is just the beginning of this process. Conducting new research and collecting clinical experience to obtain more information is necessary for learning more about augmenting communication for children and adults with JNCL, and thereby improving their life quality.

References

- Budiyanto, K., Sheehy, H. K., & Khofidotur R. (2018). Developing Signalong Indonesia: Issues of happiness and pedagogy, training and stigmatization. *International Journal of Inclusive Education*, 22, 543–559
- Clarke, M., & Price, K. (2012). Augmentative and alternative communication for children with cerebral palsy. *Paediatrics and Child Health*, 22, 367–371.
- Clarke, M., & Wilkinson, R. (2009). The collaborative construction of non-serious episodes of interaction by non-speaking children with cerebral palsy and their peers. *Clinical Linguistics and Phonetics*, 23, 583–597.
- Downing, J., & Eichinger, J. (1990). Instructional strategies for learners with dual sensory impairments in integrated settings. *Journal of the Association for Persons with Severe Handicaps*, 15, 98–105.
- Elmerskog, B., & Fosse, P. (Eds.) (2012). *Lære for livet. En innføring i pedagogikk og læring ved juvenil neuronal ceroid lipofuscinose (JNCL)* (Learning for life. Introduction to teaching and learning in JNCL). Trondheim: Akademika.
- Gayton, R. (1982). The education and care problems of visually handicapped children suffering from juvenile JNCL. Masters thesis, University of Birmingham, UK.
- Glacken, M., Healy, D., Gilrane, U., Gowan, S. H. M., Dolan, S., Walsh-Gallagher, D., & Jennings, C. (2018). Key word signing: Parents' experiences of an unaided form of augmentative and alternative communication (Lámh). *Journal of Intellectual Disabilities*, doi.org/10.1177/1744629518790825
- Goldbart, J., & Marshall, J. (2004). «Pushes and pulls» on the parents of children who use AAC. *Augmentative and Alternative Communication*, 20, 194–208.
- Goldware, M., & Silver, M. (1998). AAC strategies for young children with visual impairment and multiple disabilities. Paper presented at the CSUN Conference, Los Angeles, CA, March 1998.
- Grini, K., & Aasen, G. (2013). *Taktile symboler og planer (Tactile symbols and plans)*. <http://www.statped.no/>
- Grove, N. (1990). Developing intelligible signs with learning-disabled students: A review of the literature and an assessment procedure. *International Journal of Language and Communication Disorders*, 25, 265–293.
- Grove, N., & Woll, B. (2017). Assessing language skills in adult key word signers with intellectual disabilities: Insights from sign linguistics. *Research in Developmental Disabilities*, 62, 174–183.
- Hendrickson, H., & McLinden, M. (1995). Study of raised symbolic communication systems for visually impaired people with additional learning difficulties. *International Journal of Language and Communication Disorders*, 30, 526–533.
- Hörmeier, I., & Renner, G. (2013). Confirming and denying in co-construction processes: A case study of an adult with cerebral palsy and two familiar partners. *Augmentative and Alternative Communication*, 29, 259–271.
- Jones, F., Pring, T., & Grove, N. (2002). Developing communication in adults with profound and multiple learning difficulties using objects of reference. *International Journal of Language and Communication Disorders*, 37, 173–184.
- Marvin, C. A., & Privratsky, A. J. (1999). After-school talk: The effects of materials sent home from preschool. *American Journal of Speech-Language Pathology*, 8, 231–240.

- McLarty, M. (1997). Putting objects of reference in context. *European Journal of Special Needs Education*, 12, 12–20.
- Millar, S. (1998). The child, education and augmentative and alternative communication. In A. Wilson, A. (Ed.), *Augmentative communication in practice* (pp. 54–61). Oxford, UK: CALL Centre.
- Millar, S., & Aitken, S. (2003). *Personal communication passports: Guidelines for good practice*. Oxford: CALL Centre.
- Park, K. (1995). Using objects of reference: A review of the literature. *European Journal of Special Needs Education*, 10, 40–46.
- Park, K. (1997). How do objects become objects of reference? A review of the literature on objects of reference and a proposed model for the use of objects in communication. *British Journal of Special Education*, 24, 108–114.
- Roche, L., Sigafoos, J., Lancioni, G. E., O'Reilly, M. F., Green, V. A., Sutherland, D., van der Meer, L., Schlosser, R. W., Marschik, P. B., & Edrisinha, C. D. (2014). Tangible symbols as an AAC option for individuals with developmental disabilities: A systematic review of intervention studies. *Augmentative and Alternative Communication*, 30, 28–39.
- Rowland, C., & Schweigert, P. (2000). Tangible symbols, tangible outcomes. *Augmentative and Alternative Communication*, 16, 61–78.
- Rowland, C., & Schweigert, P. (undated). *Tangible symbol systems primer*. May be downloaded from <https://www.designtolearn.com/uploaded/pdf/Tangible-Symbols-Primer-07-09x.pdf>
- Rudd, H., Grove, N., & Pring, T. (2007). Teaching productive sign modifications to children with intellectual disabilities. *Augmentative and Alternative Communication*, 23, 154–163.
- Sablitzky, J. (2009). Life with Batten disease. *Insight*, 2 (4), 39–40.
- Solomon-Rice, P., & Soto, G. (2011). Co-construction as a facilitative factor in supporting the personal narratives of children who use augmentative and alternative communication. *Communication Disorders Quarterly*, 32, 70–82.
- von Tetzchner, S. (1992). A conversational aid for a girl with Spielmeier-Vogt disease. Presented at the Fifth Biennial Conference of the International Society for Augmentative and Alternative Communication, Philadelphia, USA, August 1992.
- von Tetzchner, S. (1993). *Utviklingen hos mennesker med Spielmeier-Vogt-syndromet med spesiell vekt på språklige og kognitive funksjoner (Development in individuals with Spielmeier-Vogt syndrome with emphasis on linguistic and cognitive functioning)*. Melhus, Norway: Tambartun Resource Center for the Visually Impaired.
- von Tetzchner, S., Fosse, P., & Elmerskog, B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta*, 1832 (2013), 1894–1905.
- von Tetzchner, S., Launonen, K., Batorowicz, B., Nunes, L. R. D. O. D. P., Walter, C. C. D. F., Oxley, J., Massaro, M., Stadskleiv, K., Yang, C.-K., & Deliberato, D. (2018). Communication aid provision and use among children and adolescents developing aided communication: an international survey. *Augmentative and Alternative Communication*, 34, 79–91.
- von Tetzchner, S., & Martinsen, H. (2000). *Introduction to augmentative and alternative communication*. London, UK: Whurr/Wiley.
- von Tetzchner, S., & Stadskleiv, K. (2016). Constructing a language in alternative forms. In M. Smith & J. Murray (Eds.), *The silent partner? Language interaction and aided communication* (pp. 17–34). Guildford, UK: J&R Press.
- Woodall, J., & Charnock, D. (2017). *Tassels – Tactile learning for sensory learners, Second edition*. Hove, UK: Pavilion publishing.

Teaching Reading and Writing to Students with Juvenile Neuronal Ceroid Lipofuscinosis

*Anne-Grethe Tøssebro, Anna Christal, Deirdre McElrone,
June Cameron, Marja Leena Forssas and Tarja Eskonen*

Literacy is a basic element of modern cultures and teaching of reading and writing is a core element in education. Literacy allows information seeking and is a tool for collective memory and history construction, as well as for self-expression. Literacy is also important in communication and when using technical aids and computer programs. It is an aim that all children should be given opportunities to reach their individual learning potential, independent of their abilities and disabilities. There are teaching guides and teaching materials for braille in many countries, developed for children with visual impairment. For children with juvenile neuronal ceroid lipofuscinosis (JNCL), visual impairment and dementia will influence their ability to maintain reading, writing and mastery of literacy throughout childhood and adulthood, but their desire for self-expression may be maintained and expression through writing may remain a central activity even in adulthood.

Teaching of literacy

The function of written language is to reflect the spoken language (de Saussure, 1974; Liberman, 1999). Thus, teaching reading means helping the student understand the relation between spoken language and the characters representing the alphabet. This main aspect of literacy teaching is the same regardless of the modality of the characters to be taught, whether they are print, braille or Morse.

Literacy instruction includes two main approaches. *Skill-oriented instruction* is described as a phonological part-to-whole approach (Wormsley & D'Andrea, 1997) typically starting with speech sounds and letters, followed by spelling to construct words, or an initial whole-word approach which starts with words that are then analyzed into spelling. The teaching is generally the same, there is no individual choice, and reading materials tend to be prefabricated. *Meaning-oriented*

instruction is a whole-to-part approach, starting with meaningful sentences or text that is subsequently broken down into words and letters. The material consists of stories and key words from the children's own experiences, knowledge and interests, and focuses on both whole word and letter recognition. The reading material is often produced through a collaboration between teachers and students (Wormsley & D'Andrea, 1997; Wormsley, 2016). Many teachers use a combination of the two approaches, to accommodate the students' individual needs.

Literacy learning is often described as a stage-like process. *Emergent reading* is the first stage. The child understands that spoken words correspond to written words and subsequently that words are made up of letters and letter clusters that correspond to speech sounds. *Beginning reading* is the second stage. The child learns elementary-level reading skills and understands the contents of sentences and short and simple texts. Finally, *advanced reading* implies reading for inquiry and knowledge building (Kamei-Hannan & Ricci, 2015). Teaching literacy is therefore not only about teaching the code (Swenson, 2016; Wormsley & D'Andrea, 1997).

Modes of reading

Reading can be visual (ordinary print, alphabetic or logographic) or tactile (braille or Moon). Access to texts can also be auditory, such as when text is read aloud by another person or by text-to-speech programs with synthetic speech. Studies show that many individuals with visual impairment vary between tactile reading, visual reading and auditory text access (Vik, 2008; Vik & Fellenius, 2007). For a short period, students with JNCL may be able to use all three modes, but for many, the visual reading mode becomes inaccessible relatively early. When a child has received a JNCL diagnosis, it is assumed that the future reading mode will be tactile and that auditory text access will be very important. Listening to audio books remains a favorite activity for many adolescents and adults with JNCL (see also Chapter 19).

There are two main tactile reading systems in use by students with JNCL: Moon and braille. For a period of approximately 200 years, Moon and braille were both used, but about 50 years ago, the use of Moon subsided. One important reason was the lack of functional writing tools for Moon relative to those available for braille, which had the slate and stylus and later the Perkins Braille (see Figure 14.5 below). Today, the problems related to writing Moon letters are solved because Moon letters and text can be printed using an *embosser*. However, today braille is used all over the world while Moon is mainly used in the UK and alongside with braille.

Moon

Moon is a tactile alphabet and the shape of many Moon letters are similar to printed capital letters or parts of capital letters (see Figure 14.1). Because of the strong resemblance to the print alphabet, Moon seems easier to learn than braille for peers and people in the person's environments. The Royal National Institute of Blind People (RNIB) published a report based on interviews with teachers and other professionals working with Moon (Cryer, Home, & Wilkins, 2011). The report suggests that Moon is easier to learn than braille. Moon letters are easy to identify with touch, they have a strong resemblance to the print alphabet, and are easy for sighted people to learn (peers, parents, teachers etc.), and the use of Moon can therefore lead to more inclusion than braille. Moon can be produced in any size and can be printed on an embosser, like braille. In the UK, Moon is recommended (a) for persons who used to have normal vision but who have lost their eyesight and therefore need to learn a tactile reading mode, and (b) for students who for some reason have problems learning braille.

In the UK, Moon is chosen for some students with JNCL because most children with JNCL have achieved some print reading and writing skills before they start to lose their vision. In the present project (see Appendix A), some of the students did not become good braille readers. It is possible that Moon might have been a better alternative for some of them, as suggested by these parents' comments.

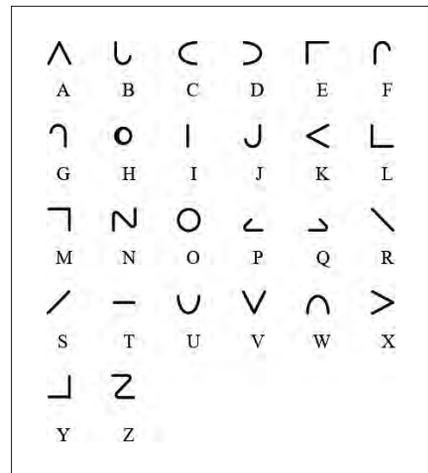


Figure 14.1. The Moon code

My daughter would do well in spelling tests up until 8, I seem to remember words like elephant and started to be less able to retain the spellings from memory. Braille was ok at first. She learned the alphabet and she started grade 2 braille but then started to forget even when enlarged and the VI teacher was quite disparaging (before diagnosis). It was a distressing time. I had researched and found Moon font and attended a conference and Moon base and it was agreed to try it and it was very successful and my daughter really enjoyed having her own language and the symbols being similar to the words she had learned.

Reads in Moon, was taught this over a period of a school year before sight had completely gone. Moon was chosen over braille as the VI teacher wanted to have something in place for when fine motor skills started to decline. Reads well in Moon although is a little slower and reads shorter books.

In the early stages of Moon it was very successful but lack of resources meant that books were mainly homemade ... the font size needed to be bigger than standard Moon font. My daughter really enjoyed Moon.

Braille

Braille is a system of raised dots. The dots in a braille cell are numbered from 1 to 6. The left column consists of the dots 1, 2, and 3 with 1 at the top, 2 in the middle and 3 at the bottom. The right column has dot number 4 at the top, 5 in the middle and 6 at the bottom. Braille letters consist of different dot combinations, for example the letter *m* consists of dots 1, 3 and 4 (see Figure 14.2).

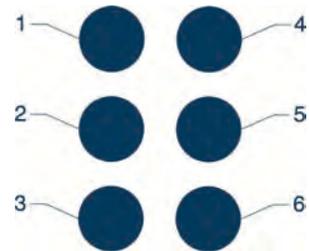


Figure 14.2 The braille cell

Braille is usually read by moving the fingers on both hands from left to right while touching the braille letters lightly and smoothly with the fingertips. To facilitate writing braille on a computer the braille cell is extended to 8 dots to make more dot combinations possible. Computer-based reading and writing tools have made it possible for braille readers to navigate on the internet and have access to synthetic speech that reads the text on the screen. Braille is the tactile alphabet that is most commonly used all over the world.

Uncontracted and contracted braille

Uncontracted braille corresponds to visual writing as all the letters in words are written, for example the word *happy* in Figure 14.3. Figure 14.3 also shows an example of a sentence written in contracted braille. Contractions are abbreviations, such as writing the letter combination *th* with one instead of two braille cells. Also, *in* is written with one braille cell in contracted braille. The word *thin* consequently is written with two braille cells in contracted braille, one braille cell for *th* and another braille cell for *in*. The word *braille* is written with three braille cells as shown in figure 14.3. There are several levels of contracted braille. Braille readers will usually achieve a higher pace when reading contracted braille.

and unambiguous as possible to facilitate literacy learning for braille readers worldwide (Cryer et al., 2013). Tactile reading is considered more difficult to learn than visual reading and represents bigger challenges for memory. Sighted children meet written words and texts in their environment to a much higher extent than children who are blind and are thus incidentally exposed to varied learning opportunities every day (McCall, 1999). To educate teachers and facilitate better learning opportunities for pupils with blindness, several countries have defined special learning areas for children with severe visual impairment. An example is the Expanded Core Curriculum (ECC) as described by Lohmeier (2009), which promotes tactile reading and writing as core components.

Traditionally, teaching of braille is mostly based on a parts-to-whole approach. This is due to the fact that tactile readers are not able to perceive whole written words or sentences simultaneously, such as sighted students do (Millar, 1997). During reading, the fingers of the tactile reader will encounter one letter at a time. In skill-oriented instruction the child learns the smallest elements first, that is, the letters, and builds upon these fundamental skills to develop more sophisticated literacy skills. This teaching method is the traditional method for teaching literacy both for visual and tactile reading and writing (Millar, 1997; Wormsley & D'Andrea, 1997).

Since the 1940s different teaching methods based on a whole-to-parts meaning-oriented instruction have been developed. These methods were not considered applicable for teaching braille because of braille readers' problems perceiving larger entities like letter combinations or words simultaneously. However, since the 1990s whole-to-parts methods have been adapted to suit braille readers. An example is I-M-ABLE (see below) that was developed specifically for at-risk students who found learning braille difficult and showed a slower learning pace than expected for their age (Wormsley, 2016). Slow learning pace can be due to for example language disorders, dyslexia, intellectual disability or attention deficit disorders.

Literacy teaching should suit the individual needs of the student in the best possible way. «The answer [to teaching reading] is not in the method; it is in the teacher. It has been repeatedly established that the best instruction results when combinations of methods are orchestrated by a teacher who decides what to do in light of children's needs» (Duffy & Hoffman, 1999, p. 11). Reading pace – number of words read correctly per minute – is often used to evaluate students' reading skills. Braille reading is usually slower than print reading, but the reading pace varies a lot among braille readers. Many achieve functional reading skills even if the reading pace is slow. Children develop a fast reading pace when they engage in varied, motivating and enjoyable reading (Swenson, 2016).

Braille courses for students, families and staff – an example from Finland

In many countries, the educational authorities offer braille courses to students, teachers and parents. In Finland, a five-day course for parents is organized by The Finnish Federation of the Visually Impaired (FFVI) twice a year. The aim is that the course participants should become familiar with braille as well as pre-braille skills. FFVI offers separate braille courses for professionals.

In Finland, braille teaching for children with visual impairment follows a three-step procedure. In the first step, the braille material is produced with extra spacing between letters, words and lines. When the child has achieved a smooth touch to the braille letters and shows progress in reading, teaching proceeds to the second step. The extra spacing between letters and words is removed, but extra space between lines is kept to make it easier for the child to follow the lines. In the third step, the extra space between lines is also removed. Each child proceeds individually through the steps, according to his or her skills and abilities.

The resource center Valteri, Center for Learning and Consulting, Onerva-unit in Jyväskylä, organizes courses for students with visual impairment in preschool age and school age. A one-week support course for students who are blind always includes different subjects and techniques to support education and functioning in everyday life, and braille training is always included in these courses.

Reading and writing for children with JNCL

Early literacy learning for children with JNCL will be influenced by two main factors related to the disease. First, visual decline and subsequent blindness will gradually compromise visual reading and make tactile reading the only reading mode available (see Chapter 4). Second, dementia will eventually influence all learning, also literacy learning. Complex skills like reading and writing are particularly vulnerable. However, problems with memory and learning typically become apparent well after the child has started school but there is considerable variation in age of onset (see Chapter 5). Also, reduced skills in the hands and arms will affect tactile reading and writing skills, but for the majority of persons with JNCL this occurs quite late in school age. In the present study, the mean age for onset of these kinds of motor problems was 15.5 years (see Chapter 7). At this age, many children with JNCL who had relevant training have been tactile readers for several years.

There is a relationship between reading and working memory, but there is not agreement between researchers with regard to how working memory influences reading and reading disorders (Kendeou, van den Broek, Helder, & Karlsson, 2014;

Nouwens, Groen, & Verhoeven, 2017; Pelegrina, Capodieci, Carretti, & Cornoldi, 2015; Pimperton & Nation, 2014; Van Dyke, Johns, & Kukona, 2014). Some authors have suggested that braille places greater demands on working memory than print (Daneman, 1988) and others that problems with working memory capacity will make it very difficult or impossible for students with JNCL to learn to read and write (Kristiansen, 1988). However, research findings contradict the claim that it is very difficult or impossible for children with JNCL to learn to read and write. A review of the reading and writing skills of 39 students with JNCL who received support from Tambartun resource center (Statped) in the period from 1995 to 2011 reported that 23 of the 39 students could read and write braille at some level, but skills varied considerably among the students. Seven of the 39 students could write but not read braille. These students used braille keyboards and displays, except for one student who touch-typed on an ordinary keyboard. The remaining nine of the 39 students were unable to read and write braille (personal communication, Mohammed Beghdadi, 2012). These findings indicate that a majority of this sample of students with JNCL benefited from tactile reading and writing, and thus support the claim that students with JNCL should be given opportunity to learn braille, although for some, learning Moon might be a better option.

Also, findings from the present survey (see Appendix A) contradict the view that students with JNCL are unable to learn to read and write. The parents were asked to indicate their child's best level of reading competence in different age periods, that is, whether the child was a) reading letters, b) reading words, c) reading short texts, or d) reading books. Both reading print and reading braille were assessed. Seventeen participants of a total of 107 (15.9%) were reading books in braille during at least one of the age periods. Two of the 17 children had good braille reading skills before the age of seven, because they had started receiving braille instruction very early. Another 21 persons (21.6%) were able or had been able to read short texts. These findings show that about one-third of the students in the project developed quite good braille reading skills. We note that some of the children in the project were rather young, and it is possible that some of them might have the capability to learn to read books or short texts when they are older.

The age of the participants ranged from 6 to 34 years. Figure 14.4 show the percentage of the participants who had been able to read and write a text in print or braille in different age periods. The results show that print was most used in the early phases and peaked around age 8–10. At this age, 40 percent of the children read text in print, after that the percentage dropped. Braille reading and writing peaked at 14–17 years and then started to drop. Nine participants between the age of seven and 13 years were reading in both print and braille. The typical picture is thus that most of the younger participants read either print or braille.

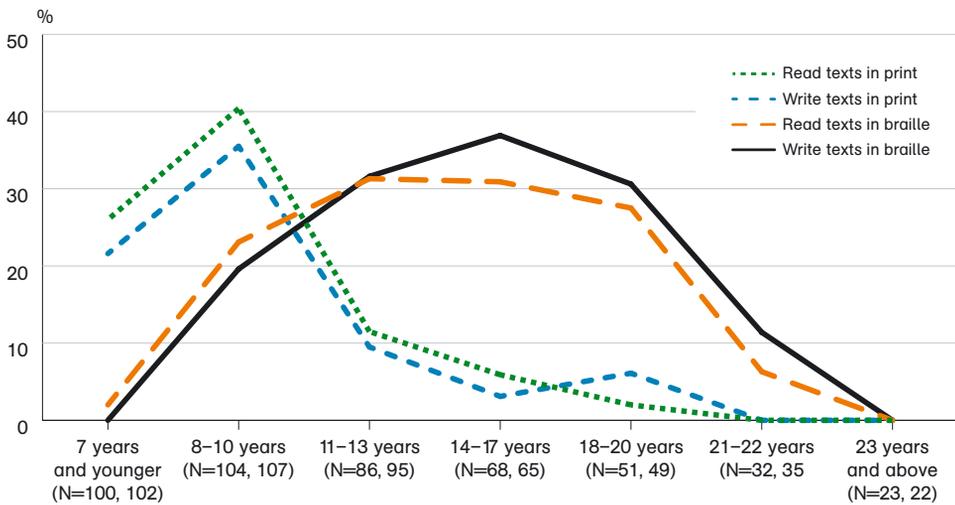


Figure 14.4 Percentage of individuals who could read and write texts in braille and print in different age periods

Note: The number of participants (N) varies due to the age distribution. Each age span has two Ns: The first is for reading, the second for writing.

Braille reading skills were further developed and maintained after the ability to read and write texts in print was lost. Figure 14.4 also shows however that ability to read short texts or books in braille did not last. Even those who developed excellent reading and writing skills eventually lost these skills as the disease progressed. The length of the period with preserved braille reading varied considerably. None of the 23 participants described by parents who had reached the age of 23 by the time of the survey practiced reading or writing, but two participants aged 21 and 22 were still reading books in braille. One person, who was only described by staff and therefore not included in Figure 14.4, was still reading at the age of 23. The findings from the present project further indicate that many adults still took pleasure in listening to audio books (see Chapter 17). Thus, the ability to enjoy the content of texts and books can be preserved through auditory access.

It seems likely that experience with reading print will facilitate braille learning. For a child who has learned to read printed letters, learning braille demands practicing the already learned reading skills with the addition of using a new modality. However, many factors may influence braille learning and it is not clear how the skills associated with reading print through a visual mode are best transferred to using a tactile reading mode. The present study indicates that children who became blind before the age of ten tended to become the best braille readers, although there were a few exceptions. Several parents mentioned that early braille learning was due to early onset of blindness:

With his rapid loss of eyesight, we began braille along with large print access. He did better with braille and really enjoyed learning it. He spent hours typing on his brailler, learning spelling words and reading his books. He was very proud of the fact he could do braille. When older he'd get frustrated with not remembering cells, the teacher just went back to what he could do and the simpler words. For him, braille was the best thing he did, he read books in the dark way after bedtime many a night.

During preschool (age 5–6) he was already interested in letters and numbers, his visual impairment increased and he suddenly lost his interest in these. Starting his second year at the school for the blind, he started learning braille and he could read like a little world champion. He also learned shorthand braille and mastered it. At the age of 20–21 years his reading competence decreased and stopped about age 22 years.

"Arthur" was blind at age six years. After that he read braille. Soon after getting blind, Arthur received teaching in braille and he was skilled in braille. However in the final stage of life, it became more difficult.

Several parents however, described late onset of braille teaching because the school was giving priority to residual vision and print reading as long as possible, such as these parents:

He stopped reading normal print, as even the screen-reading device wasn't sufficient in its magnification. Learned braille for only a short time, as his mental deterioration happened very quickly. Nevertheless, he went on practicing on type writer.

Gone blind at the age of about eight. Cognitive skills were too low from 8–10 years of age to learn braille.

Stopped reading because of cognitive decline and complete loss of sight.

Because of her blindness she cannot read normal print any longer. While it was still possible, they did not work with braille at the school for the blind. Eventually it was too late.

She wasn't able to read black print at the age of ten, neither with technical aids. Her cognitive capacities were no longer sufficient to enable her to learn braille.

Staff members also commented on braille reading:

Could read print well until JNCL symptoms at about age eight. Print was enlarged but by age 13, she could no longer read even enlarged print. Was taught grade 1 braille but progression of JNCL prevented consolidation of braille learning due to deterioration of memory.

Even in this area there was considerable variation. A few students who became blind in their teens still became good braille readers. The tendency however was that early introduction to braille appears to provide the best chance of becoming a good braille reader, irrespective of residual vision or reading skills in print. It is not possible to infer from the results whether print reading skills facilitate braille learning, but as long as the child enjoys reading print, this activity should be encouraged together with the provision of braille instruction.

The question has been raised whether students with JNCL should advance to learning how to use contracted braille. This progression would entail learning more braille symbols and abbreviations (see Figure 14.3) and learning to read words written in different ways from what they had first learned, that is, through uncontracted braille. One parent commented:

Our son began braille instruction in first grade, was fluent reading contracted braille by fourth grade and loved reading volumes of books until he was around 17. At that point fine motor coordination was declining and short passages and much younger level of texts were his favorites. He stopped reading braille at the age of 20–21.

This child had started to learn braille in first grade and became a very good reader. Literacy learning from young age might be one reason accounting for fluent reading through both uncontracted and contracted braille. Because contracted braille presents additional cognitive challenges, many children with JNCL will likely have problems learning it. Thus, for them uncontracted braille will be the most efficient way of reading. Learning uncontracted braille constitutes, in itself, a success for a child with JNCL. Teachers and families should take into account individual learning potential and motivation when deciding about which path to follow; both paths begin with uncontracted braille and the choice is then either to remain with that system, or progress to contracted braille.

Braille education for students with JNCL

In the present project, most of the participants had been offered braille instruction. However, seventeen percent of the participants did not get this opportunity. Failure to offer the opportunity of learning a tactile reading mode might be driven by parents' and teachers' low expectations for learning capacity in individuals with JNCL:

The parent thinks that the student should not use energy on something that would decline anyway. There were enough defeats.

Because of learning disorders, the professionals recommended not to start teaching braille at all. After she lost her vision all reading is impossible.

The special school would not offer braille.

Moreover, comments indicate that some schools did not focus on literacy for students with JNCL, like this teacher:

No, that [braille] is not something we are focusing on. Because those who know more about the disease have told us that she will never become a braille reader.

In an interview, a parent seemed to express regret with the decision not to teach braille:

She would have been able to learn braille, I actually think so. If we had started at once I actually think she could have learned braille.

Staff and parents alike described problems with access to professionals with braille competence when the child was not attending a special school or unit for students with visual impairments. Changes in staff resulted in loss of competence, and this usually happened several times over the school years. Transition to a new school or new counseling services could also result in long periods with no braille competence in the staff. One parent commented:

Changing teachers has led to several very long periods with no braille teaching. Knowledge and skills are lost as a result of teachers lacking competence.

A staff working with a young girl with JNCL commented:

The teacher [at the previous school] meant that they could not [learn braille]. The teachers had no competence in braille. So I said neither do I, but then you have to get somebody else come and help her.

Some parents and staff in the present project describe students with JNCL who had received braille training but who nevertheless did not learn to read or write braille and so eventually the teaching was terminated. Comments from parents indicate that this abandonment of braille instruction was related to the students' cognitive problems or lack of motivation.

He was never interested in reading.

His intellectual stagnation made it impossible for him to learn braille.

She learned to read just like most kids but at age eight she started to go blind and the school tried to work with braille. However, she never really took to braille so it made it hard to work with her. She just couldn't get it and make sense of the braille.

Dementia is one likely cause for the problems with learning braille. If learning is difficult and progress slow, problems with motivation are also likely to occur. Another factor is that some children in the general population have persistent problems learning literacy. It seems reasonable to assume that some children with JNCL have similar problems and that these problems are not specific to braille. The last quotation above illustrates the case of a child who had successfully learned to read through the visual mode (i.e., print) and experienced reading problems only with the tactile mode (i.e., braille). This case suggests the possibility of modality-specific literacy learning difficulty.

Braille literacy and impact on life

In spite of difficulties and decline in reading and writing skills, for some of the participants in the present study, literacy skills have played a crucial role in their lives, as evident in this quote from an interview with the mother of young woman with JNCL:

I think maybe the most important for her was that she took pleasure in writing when she learned writing braille. And it meant so much to her that she could write down her thoughts on paper. That was also important. She expressed it [the thoughts] that way, both pleasures (...) and when she was angry we heard her go upstairs and then we heard the Perkins brailier. But she wrote, maybe you know that, that she published a poetry collection. She wrote those things before she was 18 years old.

A staff member working with a young man recounted:

Yes, well, when he came here at the age of 23, he read braille. So he read books himself, sat by the computer.

Then [at the age of 23] braille books were sent every month from the national organization for blind people.

Today he does not read braille. That is, it happens, we have cards with braille, he plays cards, and even that is getting difficult, and that is only one letter per card.

In summary, the results from the present project show a great variety in braille reading skills among students with JNCL, which in part may be due to differences in age when teaching of braille was initiated. Some of the participants became proficient readers and some did not progress beyond letter- or single-word recognition. The participants' interest in reading and writing also varied considerably. Some took great pleasure in reading or writing, and some did not. Some were never given the chance to become literate as braille teaching was never offered. Some of the quotations above are striking in that they speak to a strong liking for self-expression of one's own thoughts, ideas, feelings through the medium of written language, an interest that was maintained over many years. Braille made this self-expression possible.

Strategies in teaching reading and writing skills

Reading and writing are both technical and functional skills. Technical skills are about recognizing letters and words, producing letters and spelling words. Functional skills concern the use of reading and writing for a purpose, to do something with words, such as writing a story, doing school work, reading a text to find specific information, or reading the day schedule. It is important to ensure

that the child's reading and writing become functional from the very beginning. Children with visual impairment should encounter tactile writing everywhere in the environment, just as sighted peers experience visual writing.

Reading and writing braille

Teaching braille usually starts with teaching letter and speech-sound correspondence, and print-braille correspondences if the child has already learned to read print, and then proceeds with words and sentences. Teaching starts with the letters that are easiest to recognize and discriminate. Many children with JNCL develop an interest in reading early and find reading rewarding because it gives access to literature and information. However, some children find learning to read and write very difficult. Their progress is slow, they feel unsuccessful and may lose the motivation to learn to read and write.

Most students with visual impairment start learning to write braille on a Perkins brailler, which has six keys, a space bar, a line spacer and a backspace (Figure 14.5). The six main keys correspond to the six dots of the braille cell. Paper is inserted into the top of the brailler and once the keys are pressed, raised dots appear on the paper. During writing the six writing fingers are permanently placed on the six keys and there is no need for searching to locate letters.

There are also braille keyboards for computers (see Figure 14.6). When writing on a computer the student can receive continuous feedback with synthetic speech, which makes it easy to discover and correct spelling mistakes. The keyboard in Figure 14.6 has a tactile display of 40 braille cells so the student can read the last written words with the fingers.

Children who have started to write on a Perkins brailler later usually switch to a braille keyboard and synthetic speech. Pressing the keys on a Perkins brailler requires more strength than pressing the keys on a braille computer keyboard. Using a Perkins brailler thus has positive effects for finger coordination and strength.

Screen readers read the information on the computer screen in synthetic speech. The user utilizes keyboard commands for navigation instead of a mouse. A screen reader can be set to read all text on the screen, including menus and dialogue boxes. Selection of a screen reader may depend on the



Figure 14.5 A Perkins brailler



Figure 14.6 A braille keyboard with a small reading display

operating system of the computer, funding and individual preference. The user can choose synthetic speech with a familiar dialect and the output speed of the screen reader, and decide which punctuations in the text will be read out loud.

Touch typing

When writing fluently on a Perkins braille or braille keyboard many students with visual impairment go on to learn touch typing on an ordinary QWERTY keyboard to increase the speed of writing. In the present project, one of the participants had learned and used touch typing.

"Jane" was fluent in reading braille and read complex language (the whole Harry Potter series, Romeo and Juliet) in braille until she was 15 years old, then lost the skill. She could produce braille, but never as well as she could read. She could type by touch, but never as well as she could read. She listened to audio books and her comprehension was good until around age 14–15. Then she started to regress, preferring books for younger children and more repetition.

The story of Jane above illustrates that some students with JNCL can learn touch typing. However, the decline in cognitive and fine motor skills will for most students compromise learning a new way of writing that is completely different from the way they already have learned. Although there might be exceptions, in general students with JNCL might benefit most from continuing to write on a Perkins braille or a braille keyboard.

Writing with Lego blocks

This is a writing tool created especially for young students with visual impairment. It consists of a Lego board and Lego blocks with braille letters (Figure 14.7). The board has six Lego blocks with each letter (29 letters in the Norwegian alphabet) and six punctuation signs, for example full stop and exclamation mark. The student can write words and sentences by placing Lego blocks with braille letters on two lines.

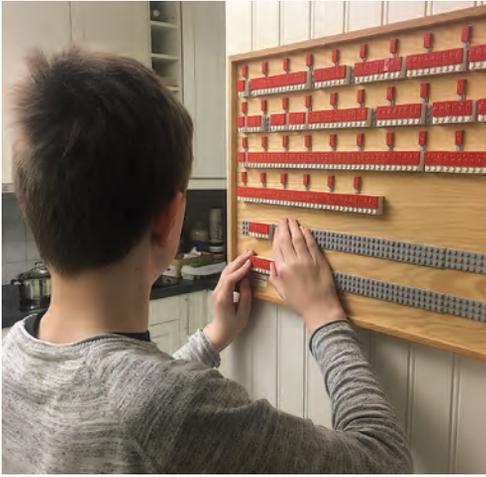


Figure 14.7 Lego board with braille letters and two lines for writing words

Reading and fingertip sensitivity

The fingertips are used when reading braille and finger-tip sensitivity is therefore important for perceiving braille cells and dots when moving the fingers across the paper. The same kind of tactile sensitivity is not required for braille writing. Writing braille is therefore considered easier to learn than reading braille, although there are individual differences even here.

Some students with JNCL never learn tactile reading but may still learn braille writing. In

fact, writing may become a person's favorite activity even when his reading skills are poor. When a person can write but is unable to read braille, it is important to ensure that the person receives reading help or has access to a computer with synthetic speech output. Lack of equipment or support may reduce the continued use of literacy skills and hinder access to an engaging activity, as indicated by the staff member's comment about the young woman below.

She became an expert on that [writing braille], she never read braille because she did not have enough motivation, but she wrote and took pleasure in writing throughout school.

I think she could have managed this [to write] even longer, but then she stopped going to school (...) and it became difficult to follow up because residential staff did not have enough competence to use the computer.

Single word reading and writing

Functional use of reading and writing skills is possible even if the person reads or writes only single words. Single-word reading or writing may be used for labeling books and teaching materials, contents of shelves and drawers, electrical equipment for cooking, *on* and *off* on light and other switches, calendars and daily or weekly schedules, or making recipes, and thus contribute to independence and orientating in the environment (Wormsley, 2003).

If a student can recognize only a few single letters, this skill may be exploited to provide more functionality. For example, people routinely use abbreviations (even single letters) as codes in text messaging to save time and effort. By pairing a single letter with an associated word, preferably a word beginning with that letter, the word's referent can be identified. For example, the braille letter *s* can indicate the activity "swimming" on the child's schedule. In some activities, single letters can be more practical than complete words because of limited writing space, such as labels on a deck of cards or other game equipment (see Chapter 21).

The I-M-ABLE method for teaching braille

I-M-ABLE stands for «individualized meaning-centered approach to braille literacy education» and is developed for emergent and beginning braille readers who have problems learning braille the traditional way and need a more individualized approach for learning tactile literacy (Wormsley, 2016). This approach was inspired by meaning-centered literacy teaching for sighted students and adapted for tactile reading on the basis of Wormsley's own experience as a braille teacher and counselor.

An important element in Wormsley's method is using words and texts that give an immediate sense of meaning for the individual, that is, based on individual experiences and interests. Wormsley claims that the students' emotional evaluation of the reading material can be of great importance. She recommends a highly individualized reading vocabulary and the production of individual teaching materials. There are no ready-made materials and the teacher creates the teaching materials after assessing the student's interests and experiences.

I-M-ABLE can be used as an alternative or a supplement to other teaching strategies. The list below contains some of the main elements in early literacy instruction from I-M-ABLE. Note that some of the elements on the list are used in both skill-centered and meaning-centered approaches.

- Initial assessment of the student's reading and writing skills and attitudes toward reading and writing.
- Collecting information about the student's interests.
- Creating a braille-rich environment: Wherever there is print, there should be braille.
- Frequent instruction. Emergent and beginning braille readers should have 1–2 hours of training every day.
- Identifying key vocabulary words and phrases. Words and phrases must be emotionally laden and promote the student's engagement in

reading activities. The key vocabulary is the basis for choosing words for the first reading instruction.

- Using brailled flash cards for the first words and phrases.
- Selecting first words and phrases that have features that make them easy to recognize and to discriminate, such as short words and long words, or different initial letters.
- Teaching tracking of finger movements to promote fluent reading and prevent unnecessary stopping and scrubbing up and down on the letters to identify them.
- Not asking the students to identify words that they do not yet easily recognize. In the early stages of reading instruction, this will probably lead to scrubbing. Tell the students what word they are going to find on the card and ask them to find the word and read it.
- Teaching reading some key words first, and then introduce the first letter.
- Making sure the teaching materials are motivating and based on individual preferences.
- Using word games as a supplement.
- Keeping continuous records of words and letters learned.

Writing stories based on own experiences and preferences

Instead of or as a supplement to ordinary textbooks, students can write their own stories. The contents and complexity will vary but the stories should be based on the student's own interests and preferences. For emergent writers, texts should be very simple and if possible contain many repetitions of words, such as in this example:

I like pizza
I like birthdays
I like Harry Potter

Repetitions may function as formulas (Tomasello, 2003) and make the story easy to read. The child may learn the story by heart and have a feeling of reading it, even if the reading skills are not good enough for spelling and reading the words. Moreover, motivation for reading one's own texts is often higher than for reading ready-made stories and texts. Motivation is very important for emergent learners, and the importance of motivation for people with JNCL has been amply demonstrated in the quotations presented earlier in the chapter.

Creating a braille-rich environment

Most students with JNCL attend mainstream classes during their first years in school. In some countries there are no or very few special schools for visually impaired children and other special needs. In such countries, many students with JNCL attend mainstream schools throughout the school years. The teachers will rely on courses and guidance from resource centers to build the necessary competence. However, when counselors visit such schools, there is often no sign of braille in the classroom, except on the desk of the student with visual impairment. There is a lot of printed materials on walls and in shelves and drawers for sighted students, but not for the braille readers who are exposed to written language only while sitting at their desk.

Braille instruction sometimes takes place in a separate room to minimize distractions for the student, and to avoid disturbance to other students from the synthetic speech or noise from the Perkins brailier. Thus, the classroom situations experienced by the braille reader and the classmates might be very different, and they may unintentionally signal that braille is not something classmates should engage in. Creating a braille-rich environment is therefore one measure to enhance inclusion in the mainstream classroom (see Table 14.1). Braille should be present in every part of the classroom, not only around the desk of the braille reader.

The classmates will also learn something about braille when braille letters and words are present in the classroom and elsewhere in the school. Braille becomes less special and more available to all students. In fact, introducing braille to the sighted classmates is the best way to ensure inclusion of the braille reader. Experience from mainstream classrooms shows that sighted students often find braille fascinating and learn braille with great enthusiasm. It is necessary that peers get some knowledge and basic skills related to braille to understand and appreciate the work of their braille-reading classmate. With rather simple means, braille can become a natural part of the school environment. Peers will become aware of the similarities between alphabets, independent of modality, so there will be learning gains for everybody.

Table 14.1 Some elements in a braille-rich environment

• Place a braille alphabet on the wall next to the print alphabet
• Write labels on the students' belongings in print and braille
• Label the contents of cabinets and shelves in print and braille
• Write messages between home and school in both print and braille
• Write lists and recipes used in the class in both print and braille
• Write song texts and other materials used for the whole class in both print and braille

Introducing braille to the classmates does not have to be time consuming. Much can be done with a couple of lessons on braille instruction, and integrating braille into some other lessons, and it may have a substantial effect on inclusion.

Simmons (1994) describes a successful approach to braille teaching in the mainstream classroom. She worked as a teacher for students with visual impairments in an elementary school, and some students in the school were braille users. However, she observed negative attitudes to braille, especially among the students who were blind. They often avoided using braille in class, such as this girl: «I saw her [a beginning braille reader] begin to develop some subtle, negative attitudes toward braille and decided I needed to act as a resource to inform and encourage developing social skills and positive attitudes toward braille». Simons therefore started to teach braille to the sighted students in collaboration with the class teacher. She gave the sighted students access to braille and tactile materials, including a Perkins brailler. The sighted students became very interested in braille and all the negative attitudes evaporated. Simons organized a voluntary braille class, which quickly grew from three students to two classes with 12 students in each. Over a period of three years, approximately 125 sighted students attended the braille classes, which were organized every day during lunch recess or just after school. Finally, Simons organized a Braille Club where sighted students and students who were blind worked together on projects with themes varying from Eastern cultures to social etiquette or Abacus. The students who were blind thrived in the positive attention from the classmates and gained confidence and status. Braille became a natural and integrated part of school life for all the students.

A positive attitude to braille learning and braille materials and tools is a prerequisite for successful braille teaching. If a student is working alone with tools that the classmates do not understand or show interest in, inclusion is likely to be seriously compromised.

Creative writing and fan fiction

As writing skills become more advanced, students will be able to write with more elaboration and complexity. Creative writing and fan fiction are examples of genres that have been important for some persons with JNCL in Norway.

Creative writing

Creative writing is based on the writer's imagination. It may include different genres, such as poetry, fiction or screenplays. The creative writing process may involve developing characters and settings for the story, elaborating the story plot and so

forth. Sometimes creative writers are writing only for their own pleasure. At other times, they may want to make the texts available to others. Then the writers must have the skills necessary for making the story interesting for potential readers.

When using creative writing in education, the individual's motivation for text production is the main success factor. Teaching of orthography will be second to encouraging the creative process and the students' wish to express their thoughts and feelings. In the final stage of the writing process, the teacher can ensure that the text becomes comprehensible with a minimum of focus on spelling mistakes and other needs for correction.

The feedback from a teacher, parent or classmate is important. The teacher's feedback should be adapted to the student's writing skills and take the form of a dialogue between the student and the teacher. The feedback should motivate the student to continue working with the text, and comments and questions should be offered during the writing process and not after the student considers the text to be finished. To ensure that the student understands the feedback, the teacher's questions and comments should be specific and directly to the point. The adult should be an interested and enthusiastic supporter in the writing process, and ideally, the student should be looking forward to the feedback.

Students will often enjoy writing more when the text is related to their personal interests and favorite activities. One of the genres within creative writing is fan fiction.

Fan fiction

Fan fiction is typically written by persons who admire a specific novel, movie, television series, and so forth. Most fan fiction writers are inspired by genres like science fiction and fantasy, such as the Star Wars films or the books about Harry Potter or Lord of the rings. Their focus may for example be on the story characters, the story plot, or the setting or world where the story takes place.

It is practical for the writer to locate the story in an environment described by another author, where the setting and the characters already have been developed. It saves a lot of time and work and enables the fan fiction writer to concentrate on designing a plot and creating an interesting story.

Quite a lot of people engage in writing fan fiction. Fan fiction mostly addresses a dedicated group of readers who are familiar with the literature from which the fan fiction is derived. Fan fiction enthusiasts can communicate with other fans on a variety of web sites. They can share their manuscripts and ask for reviews from other fan fiction enthusiasts. Fan fiction is rarely published, although there are some examples of published fan fiction stories. However, this requires knowledge, careful consideration and caution regarding copyright.

Three creative writers with JNCL

Early in a child's life there is no way of knowing whether writing can become an important base for future activities. It will depend on the individual's personality, interests, strengths and opportunities. This section describes the work of three Norwegian adolescents with JNCL for whom literacy has been very important. It includes short presentations of two young authors and their books, and a more extensive presentation of a young author written by his father.

Åshild – A poet writing about her own life situation

The book *Thanks for life* (in Norwegian: *Takk for livet*) contains poems that Åshild wrote during her teenage years (Figure 14.8). The poems describe her thoughts and reactions to losing her vision and having a shortened lifespan due to JNCL. Åshild's faith in God had a strong impact on her thinking and coping, and in many of the poems she asks God for answers and reassurance in a difficult life situation. Åshild passed away some years ago.

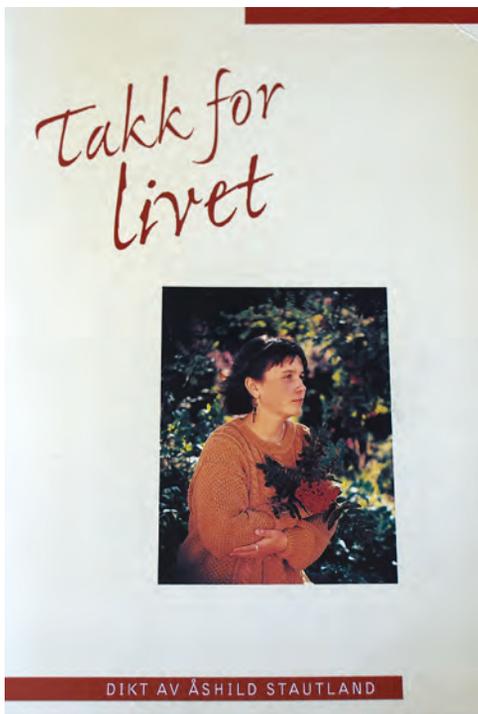


Figure 14.8 A poetry collection



Figure 14.9 A fantasy story

Ruben – Author of fantasy stories

The book *The live watermelons* (Norwegian: *De levende vannmelonene*) is about thousands of watermelons trying to destroy and take control of a city (Figure 14.9). The watermelons are stopped by the armed forces during the attempted conquest. There is a great fight between watermelons and soldiers, and as the book review stated: «You certainly don't want to miss it!».

Braille writing from a parent's perspective

This story, written by a father, integrates his son's braille writing in the wider life story of a young person with JNCL, with diagnosis, education, support services, and ordinary and not so ordinary events as part of a daily life.

When I got the message that our son had Spielmeier-Vogt disease (JNCL), it felt like walking down a hill towards a dark tunnel. I imagined myself in a spiral without the circle being closed.

Tambartun Educational Centre

We met with Tambartun, which is a state educational center (Statped) responsible for facilitating the teaching of children who lose sight – children who become blind. In life, sometimes you get a hint about something that is wise. When you are young, someone hints that she is in love with you. As an adult maybe this could be a tip about an important book, a piece of music or a bargain.

At Tambartun, a wise woman said that «maybe it is a good idea for him to learn braille while he still is using vision». That became my token.

The beginning

We started with braille training at school before the summer break at the end of the second year. Matias is seven years old. Matias learns most of the letters from the teacher. He uses a mechanical braille machine diligently every day but for short periods. The teaching strengthens the use of muscle memory, like a piano exercise. When one plays a musical instrument, one knows that this is important.

The surprise

Matias has consistently made stories that have been printed and delivered to school. Just before summer comes the surprise. He has kept a big secret. In secret, the stories are sent to Tambartun. There, the texts are made with braille lettering, and they have made a book. This book is titled «Different

Stories» and has both black writing and braille. The book becomes his reading book in fourth grade.

Introduction to braille and aids we used in the beginning

We had a 6-egg cardboard package inserted with ping-pong balls to illustrate the braille cell. We had a board with Lego – each marked with dots, like braille letters. We had a small piece of wood that showed the braille cell and a braille typewriter that prints letters on thick paper.

Autumn

A little later, in the autumn, we got computer equipment with a keyboard and a program called "Sarepta". The computer program has speech synthesis that reads letters of words and sentences. The text appears on the screen for viewing. There is a braille keyboard and a display with 80 braille cells for reading braille below. On a separate board are five buttons or keys for quick navigation. We get going.

At home, we try to write every day, preferably at the same time, with established routines. A ritual between Matias and dad is emerging. A calm, intimate gathering one-to-one, like learning to play a musical instrument. It is all about communication and little about performance. We manage five to ten minutes at a time before he is tired. We have a fixed time every day after dinner, early evening. Sometimes we only turn on the machine and then turn it off again. Other times we get some words written.

We have simple writing tasks. We try to practice the keys. It is hard to remember a sentence while remembering the letters. Matias is able to remember simple words, but not more. Dad remembers what Matias is saying and helps with the writing. The reward is that the speech synthesis reads the word and the sentence.

Eventually this develops into a conversation, where I interview Matias when he tells. When a sentence is good, I catch the sentence and he writes. During this period, I will correct the orthography without paying any attention to it.

I see progress, I see frustration, I see happiness, and I see the feeling of mastering. I expect a faster progress. It takes a much longer time. We stay focused. At the same time, he is losing his sight gradually.

Our method

We try to do the same every night.

Have as regular environment as possible.

Sometimes he does not get tired.

Sometimes we last for a long time.

Now he asks: are we not going to write? Earlier he could not remember long words, he is doing well now. Now he can complete sentences. Reading skills increase as he reads his own texts.

At school

A story is read in class, it gives self-esteem. He writes a poem.

First example text:

The stupid troll and the fox by Matias 12 years old

Once upon a time, there was a troll who had two hearts, but one of them was false. He boasted about it to a bear, and he said the bear would not be able to find it. However, the bear set off to look for it. He met an old woman who had her nose stuck in a stump. The old woman said: «Can you help me, I have had no food for a hundred years». «Well, if you have had no food for a hundred years then you can wait for a hundred years more», he said and laughed.

At home

It is easier to get started with the writing at home when there is something self-perceived to be told. Matias reflects on books we read in the evenings, or he uses stuff from movies and from fairy tales. Dad is easily bored, so he has to read something for Matias that he likes too. We read Harry Potter, Gulliver's Travels, Robinson Crusoe and Captain Nemo, Twenty Thousand Leagues Under the Sea and The Mysterious Island. We read The Three Musketeers. We read Les Miserables, and much more.

Learning

We work one-to-one. I am his supporter. I am organizing and correcting text. It is not so important to write correctly. Speech synthesis is the reward when the sentence is read. The teaching should not lead to any qualification. That is not the goal. Learning is a way to participate in society. We learn about things. About science, about history, about Christianity, religion, philosophy and philosophy of life. We learn math and we learn physics. In order to participate in the community around you, you must understand what others are talking about.

Learning = Participation in society.

Learning = not necessarily a qualification for a profession.

He calls himself an author

Matias has read all the books about Harry Potter, Hermione Granger and Ron Weasley. Afterwards he is upset. There are no more books. He decides to write a continuation of the books about what happened to the children of Harry and Ginny, Ron and Hermione. It is called "fan fiction".

He is engaged. Coming home from school, goes up to his room and writes. Together we correct the texts.

We engage his little brother to make drawings.

Father is the main text proof reader. Mother is given the role as publisher consultant and editor. She checks whether the texts are consistent. She checks that the texts have facts, relationships with people, and that the stories are in a logical development.



Illustration to one of Matias' stories

Second example text:

In the streets of London, a sailor walks on the cobblestones. He walks towards the hiring office.

«I have always enjoyed cemeteries. For some, death is an end, for me it's a beginning. I wanted a job. It was given to another person. He is going to be killed.»

Chapter 1. Outside the Sea

«In this destiny, I write Captain Landen Snow, my ship Murovania is sailing to Cuba and the sugar plantations.»

Unexpectedly somebody knocked on the door.

«Come in!» Said Landen with a tired voice.

Through the door of the door came a tall man with a sailor hat, an oilskin mackintosh and long boots.

«Captain, I have a message for you. We have sailed straight into a shear. We could not stop when we heard the foghorn.»

Third example text:

Fairytale three frogs

Once upon a time there were three frogs. They lived in a swamp. They had a good time there. A troll too lived in the swamp. The troll was a moody lazy toad, ugly as a tree and grumpy as an old woman. The frogs were in fact three princes who had been bewitched after they had eaten fungi. The only thing which could make them princes again was an egg cracked by a dragon high up in a large mountain far up in the sky.

Ashlad from the Trysil wood in Alvdal came to search for a girlfriend. He met the three frogs. They told him that they had been bewitched and that the only thing which could save them, was an egg cracked by a dragon. Ashlad went to search for the dragon. After a while he met an old man. The man asked whether he could please have some food. I have not eaten in a hundred days. Yes, but I only have an oatcake, and a drop of flat beer.

Status today

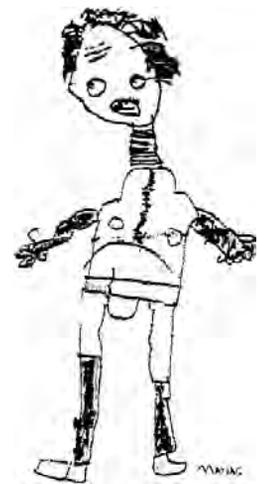
Matias is completely blind. He will have his 23-year birthday in the summer. He walks with his stick and a companion. He is hard to understand when he talks.

Matias writes a lexicon about Harry Potter from A to Z. The letters A to D he wrote himself. Now mom is a consultant. Together they go online to find facts. They have an intimate gathering. Now the mother does the typing. Matias participates with his knowledge. It is nice for Mom and son, the moments when they write together.

Grandmother is also engaged to correct texts with short stories he has written. When Matias visits grandmother, they read the text together. This makes



Self-portrait



Portrait of a man in the future

communication much easier. The time they have together means a lot to grandmother. She likes that they are engaged in something he makes.

Why strive to learn?

Matias learned to write braille by slowly developing in a safe environment, spending time exercising and practicing. Learning goals that his father thought would be achieved in two to three weeks took maybe half a year. However, as the time went by, learning became fast, the texts became better and longer. I have been his supporter from the start. After a while, I just needed to be nearby, outside his writing desk. He managed the rest himself.

As told, learning is a way to participate in society. Our goal is not a qualification for a profession. Learning gives the feeling of mastering something. It is important for participation to be able to share things with others.

For Matias, it has meant a lot to be able to express himself in writing. He has been able to express his creative abilities for a long time. He has a feeling of mastering; he has become a happy and harmonious boy. For a long time it was both reading and writing. Now he is most keen on writing the stories.

It has helped us communicating with him now that his speech is harder to understand.

Giving priority to literacy

Literacy should have high priority for all children, also for children with JNCL. For children with JNCL early intervention is important for literacy development. Teaching of braille for children with JNCL should start early (see Chapter 12).

Precautionary learning

At the time of diagnosis, many children with JNCL are still able to read print and have no immediate need for braille. Teachers should all the same start braille training to make the best possible foundation for future literacy. Otherwise, cognitive decline may make learning braille very difficult or even impossible. Braille teaching should start early to prepare for future needs.

Hastened learning

Reading and writing are complex skills and should therefore be given high priority when the diagnosis of JNCL has been confirmed. With age, learning will proceed at a gradually slower pace. To utilize the child's best learning capacity, there should be formal and informal teaching situations every day and acquired literacy skills should be useful and functional elements in the student's everyday life.

The learning environment

Some causes of poor literacy skills might be found in the educational systems. It is necessary to evaluate whether the services and the individual adaptations are sufficient, or if more should be done. It may be easy to attribute all lack of progress to the student or the disease. One should rather ask if more could be done to compensate for the loss of sight and cognitive decline, and if the teaching strategies are suited to the child's needs. One should also ask if braille teaching is starting early enough, that is, before the onset of dementia or while dementia still is in a mild phase, if the teaching of literacy is given sufficient priority and if the student is provided with varied reading and writing opportunities every school day. Comments from a staff member demonstrate the need for asking about these issues:

Earlier he received teaching in braille. He did however not benefit from this teaching because braille was very difficult for him to learn. Maybe it was presented too late.

One of the questions in the present survey asked about the usefulness of different school subjects in situations outside or after school. Some of the parents' comments concerned reading and writing.

Writing/reading: When school ended, his vision and fine motor skills had deteriorated to a degree where he was no longer able to use them. All the same they were of value when he still had these abilities.

She remembered very well the things that were important to her well past the age of 20. (...) She mastered typing on a computer with a speech synthesizer and wrote even at age 22, but at that age she lost the ability to form words by writing. When I told her letters she was able to write them on her computer.

Writing, reading and music were important and made her life rich. She enjoyed writing stories and poems already in primary school. The poems (...) often came in periods when she was frustrated by her life situation. I collected and took good care of her poems. They were her products, something she had created.

References

- Argyropoulos, V. S., & Martos, A. C. (2006). Braille literacy skills: An analysis of the concept of spelling. *Journal of Visual Impairment and Blindness*, 100, 676–686.
- Cryer, H., Home, S., & Wilkins, S. M. (2013). Unified English Braille in the United Kingdom: Part 2 – Examination by literacy braille users, braille teachers, and transcribers. *The British Journal of Visual Impairment*, 31, 238–247.
- Cryer, H., Home, S., & Wilkins, S. M. (2011). *User requirements for Moon: An informal investigation with experts*. Birmingham, UK: RNIB Centre for Accessible Information.
- D’Andrea, F. M., & Swenson, A. (2002). *Instructional strategies for braille literacy*. New York: AFB Press.
- de Saussure, F. (1974). *Course in general linguistics*. Glasgow: Collins.
- Duffy, G. G., & Hoffman, J. V. (1999). In pursuit of an illusion: The flawed search for a perfect method. *The Reading Teacher*, 53, 10–16.
- Kamei-Hannan, C., & Ricci, L. A. (2015). *Reading connections: Strategies for teaching students with visual impairments*. New York: AFB Press.
- Kendeou, P., Van Den Broek, P., Helder, A., & Karlsson, J. (2014). A cognitive view of reading comprehension: Implications for reading difficulties. *Learning Disabilities Research and Practice*, 29, 10–16.
- Kristiansen, E. (1988). Psychological/pedagogical treatment versus medical treatment. In P. Uvebrant & H. Svensson (Eds.), *Proceedings of the Nordic Meeting on Spielmeier-Vogt’s disease* (pp. 29–36). Almåsa: The Swedish Federation of the Visually Handicapped.
- Liberman, A. M. (1999). The reading researcher and the reading teacher need the right theory of speech. *Scientific Studies of Reading*, 3, 95–111.
- Lohmeier, K., Blankenship, K., & Hatlen, P. (2009). Expanded core curriculum: 12 years after. *Journal of Visual Impairment and Blindness*, 103, 103–112.
- McCall, S. (1999). Accessing the curriculum. In C. Arter, L. H. Mason, S. McCall & S. Stone (Eds.), *Children with visual impairment in mainstream settings* (pp. 29–40). London: David Fulton.
- Millar, S. (1997). *Reading by touch*. London, UK: Routledge.
- Nouwens, S., Groen, M. A., & Verhoeven, L. (2017). How working memory relates to children’s reading comprehension: The importance of domain-specificity in storage and processing. *Reading and Writing*, 30, 105–120.
- Pelegrina, S., Capodici, A., Carretti, B., & Cornoldi, C. (2015). Magnitude representation and working memory updating in children with arithmetic and reading comprehension disabilities. *Journal of Learning Disabilities*, 48, 658–668.

- Pimperton, H., & Nation, K. (2014). Poor comprehenders in the classroom: Teacher ratings of behavior in children with poor reading comprehension and its relationship with individual differences in working memory. *Journal of Learning Disabilities, 47*, 199–207.
- Sablitzky, J. (2009) Life with Batten disease. *Insight, 24*, 39–40
- Simons, B. (1994). How to make a braille wave. Paper presented at the meeting of the Association for Education and Rehabilitation of the Blind and Visually Impaired, Dallas, Texas.
- Swenson, A. M. (2016). *Beginning with braille. Firsthand experiences with a balanced approach to literacy*. New York: AFB Press.
- van Dyke, J. A., Johns, C. L., & Kukona, A. (2014). Low working memory capacity is only spuriously related to poor reading comprehension. *Cognition, 131*, 373–403.
- Vik, A. K. (2008). Individual and environmental conditions for the literacy development of students with severe visual impairments. *Journal of Assistive Technologies, 2*, 4–15.
- Vik, A. K., & Fellenius, K. (2007). Coping strategies in reading: Multi-readers in the Norwegian general education system. *Journal of Visual Impairment and Blindness, 10*, 545–556.
- von Tetzchner, S., Fosse, P., & Elmerskog B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta, 1832*, 1894–1905.
- Wormsley, D. P. (2003). *Braille literacy. A functional approach*. New York: AFB Press.
- Wormsley, D. P. (2016). *I-M-ABLE: Individualized meaning-centered approach to braille literacy education*. New York: AFB Press.
- Wormsley, D. P., & D'Andrea, F. M. (Eds.) (1997). *Instructional strategies for braille literacy*. New York: AFB Press.

Physical Education and Activity for Individuals with Juvenile Neuronal Ceroid Lipofuscinosis

*Bengt Elmerskog, Svein Rokne, Bjørg Stenersen,
Rita Jeremiassen and Riikka Hokkanen*

Motor and physical activities are important for promoting health for people of all ages, and regular physical activity is one of the most important activities people can do for sustaining best possible health (Donnelly et al., 2016; Janssen & LeBlanc, 2010; World Health Organization, 2018). The course of juvenile neuronal ceroid lipofuscinosis (JNCL) involves motor impairments, which are likely to influence both somatic health and social participation. Persons with JNCL usually show problems with posture, gait and motor coordination resembling symptoms of Parkinson disease. Hypokinesia and rigidity of muscles and joints lead to difficulty performing routine activities, such as moving around, exploring the surroundings, walking to school or shopping. Similarly, fine motor skills may deteriorate, resulting in difficulty dressing, switching on a computer, packing a school bag, making a sandwich or tying shoelaces. The gross motor problems generally become noticeable around the age of 10-to-12 years, but there is significant variation (see Chapter 7). The aim of physical intervention is to prevent and postpone the problems, and intervention must be based on knowledge about motor disabilities in general and JNCL in particular (Jeremiassen, 2016). The present chapter discusses the importance to individuals with JNCL of physical activity in general, participation in activities involving physical actions, and physiotherapy and other interventions targeting physical engagement in activities of daily living.

Physical activity and JNCL

In their early years, most children with JNCL have similar interests and participate in the same activities as their peers and show normal physical development. By the age of six, they have learned many basic skills, such as walking, running and jumping. Similarly to other children, they view themselves in terms of

their potential for action, such as being or becoming football players, swimmers, climbers, or skiers. The situation changes when the visual loss starts to affect their ability to move around freely. Participation in activities like football, tree climbing or running becomes difficult when vision is severely reduced or lost. Still, many individuals with JNCL maintain an interest in physical activities like sports, dancing and outdoor life into adolescence, even if the activity level has declined considerably. The visual impairment will however affect the child or young person's agility, speed and balance, and usually result in a more sedentary lifestyle characterized by less participation in activities involving motor skills. There is therefore a need for support and adaptation to compensate for physical inactivity caused by visual impairment (see Chapter 4 and 7). The decline in motor performance will aggravate the situation and make further development of motor skills more difficult. Stooped posture and poor balance are common in individuals with JNCL from adolescence, and many rely on wheelchairs for mobility from late teens.

In general, motor impairments and a sedentary lifestyle will have a negative influence on physical health, cognition and academic performance, whereas physical activity may actually delay cognitive decline and protect against mood disorders (see Chapter 7). Anxiety and depression are common in adolescence, and young people with JNCL are more vulnerable to these disorders than their peers (see Chapter 27). Establishing and maintaining a healthy physical lifestyle is an important element of interventions to support positive development, learning and well-being. However, the need for a healthy physical lifestyle in children, adolescents and young adults with JNCL seems to receive little attention from those who are responsible for planning and has been neglected in research.

Physical activities among individuals with JNCL

The parents in the present survey (Appendix A) were asked how often their children had been engaged in physical activities involving gross motor actions at different ages. Figure 15.1 shows a steep decline between age seven and age 23 in the percentage of participants engaged in daily physical activities, from 74 to 26 percent. The results suggest that engagement in daily physical activities are replaced by weekly physical activities. In addition, at age 23, quite a large percentage of participants were no longer engaged in any type of physical activity. Considering the potential positive impact of physical activity, the descending curve in Figure 15.1 appears worrying.

One might accept the reported decline in physical activities as a natural consequence of blindness, dementia and motor impairment, but this stance

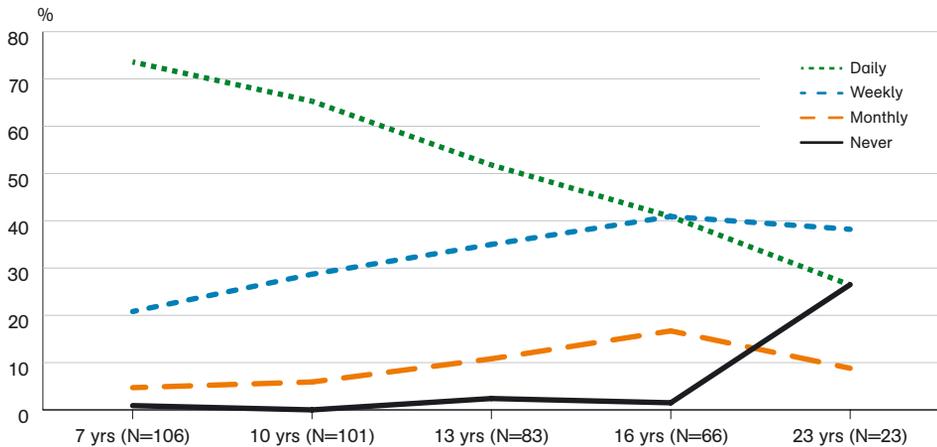


Figure 15.1 Reported frequency of physical activity at different age levels

might lead to a lack of initiative on the part of professionals. Instead one might try to identify and eliminate shortcomings in the environment by asking basic questions, such as what could be done to compensate for the lack of football participation when severe visual impairment makes playing impossible, or to compensate for difficulties in walking to school when gross motor problems become evident. Many barriers in the environment can be met by compensatory measures (see Chapter 16). Compensatory measures may include adaptation of current activities, introduction of new activities, use of technical aids, provision of support or adaptation of the physical surroundings.

The results of the present study show that physical inactivity is not inevitable for everyone. Some young adults with JNCL participate in one or more exercise programs daily, whereas some of their peers are not participating in any kind of physical exercise. Such differences may be a reflection of not only differences inherent among individuals with JNCL but also in their environment. The parents in the present study were asked to indicate who took the most responsibility for ensuring that their child with JNCL was engaged in physical activities: the person with JNCL (self-governed), the family (family-governed), the school or residence staff, or collaboration between the family, the school and the residence. Parents reported that, for seven-year olds, the responsibility was split between the children themselves (36%) and collaboration between school and family (48%). Self-governed activities were reduced to three percent at age 13. At this age, participation in physical activities was mainly family-governed for 26 percent of the participants and a result of collaboration between the school and the family for 55 percent. At 16 years the pattern had changed again: upholding a physical

lifestyle had become family-governed for 59 percent and a collaboration with others for 14 percent. This trend was strengthened for participants who were 22 years old.

The results indicate that many individuals with JNCL were engaged in physical activities on their own initiative when they were younger. However, owing to the visual impairment and emerging cognitive and motor declines the children and young persons with JNCL became more and more dependent on others in order to be engaged in physical activities. The results emphasize the importance of the collaboration between the family and the school for maintaining the young person's participation in physical activities throughout the school years. However, they also indicate that the school could have a stronger role in ensuring a physically active lifestyle for older students with JNCL. In adulthood, physical activity depends more on the residential staff. The declining curve in Figure 15.1 may indicate a need for more support and adaptations with increasing age, to support maintenance of a best possible physical lifestyle.

Physical education and physiotherapy

The parents were further asked about the impact on daily life of the physical education their child had received at school. Figure 15.2 shows that about 77 percent of the parents found that the physical education had a high or very high impact on their child's daily life. Nearly 17 percent reported a moderate impact and about seven percent found little or no impact of the physical education.

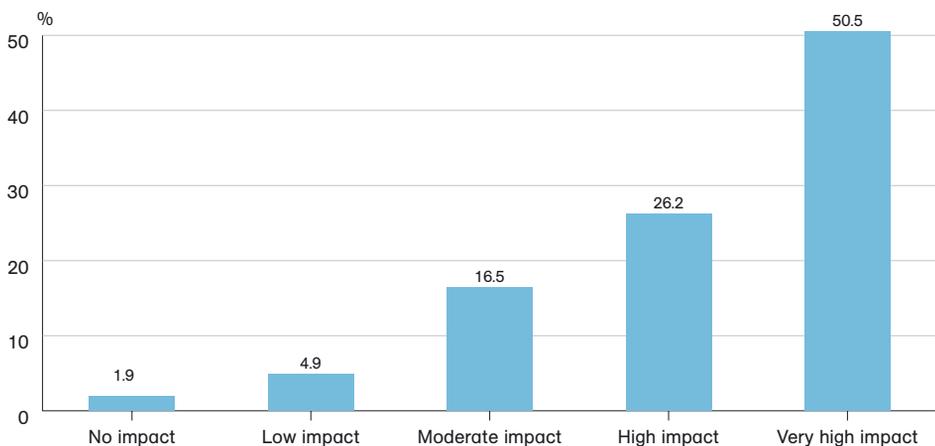


Figure 15.2 The percentage of parents giving each score for the impact of physical education on daily living (N=103)

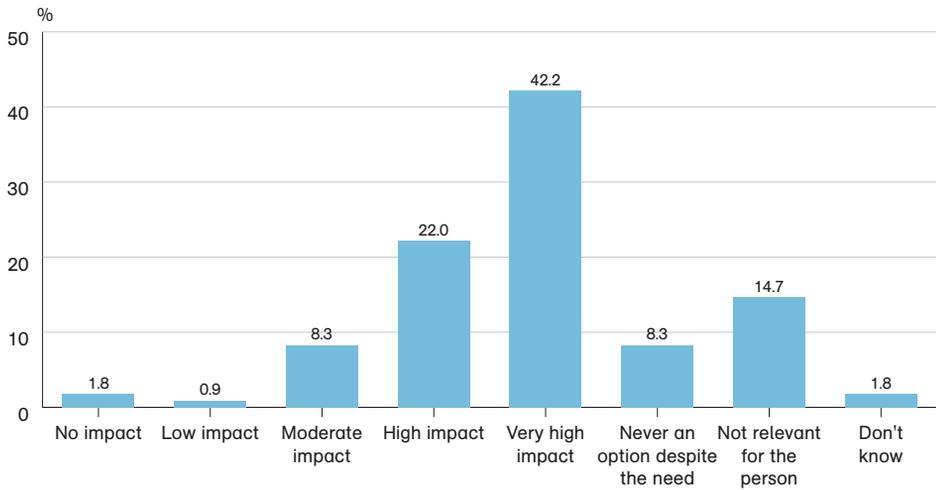


Figure 15.3 The percentage of parents giving each score for the impact of physiotherapy on daily living (N=109)

The parents were also asked if their child had been given physiotherapy and if the physiotherapy had had any positive impact on the child in daily life. Seventy-three percent of the participants had received physiotherapy, eight percent had not received physiotherapy despite a need, and for 15 percent physiotherapy had not been considered relevant (Figure 15.3). For the participants who had received physiotherapy, eighty-five percent reported a high or very high impact on their child's daily life. Eleven percent found the impact moderate and about four percent that it had little or no impact.

Physical activity in school and physiotherapy were thus evaluated as having a positive impact on the daily life of the participants. However, only a minority of the children and young people with JNCL were provided with extra physical training and some parents reported that their child had been denied physical education at school because of the disabilities.

Observations or registrations of declines and disorders are often used as the criterion for the provision of services and resources. Physiotherapy was mainly introduced as a result of physical decline. The fact that only thirty-four percent of the 7- to 10-year-olds received physiotherapy suggests that gatekeepers of services might not have viewed physiotherapy as a preventive or proactive measure for a majority of the children with JNCL in this age group. Some parents reported that they had to fight the system to obtain services, as reflected in the following comment:

Physiotherapy was finally provided, after many years of fighting, when our daughter lost the ability to walk.

Further, some parents reported that physiotherapy was terminated when the authorities or the physiotherapist could not see any evidence of positive effects of the physiotherapy:

We wanted more physiotherapy for our child, but the services were terminated when she was 18 years of age. They claimed it had no positive effect for our child

Policy barriers, such as dismissal from services when progress cannot be documented, reflect a model of service delivery that is not in keeping with the contemporary views on habilitation reflected in International Classification of Functioning, Disability and Health (Castro & Palikara, 2018). The implications of a failure to consider preservation of existing functionality and prevention of unnecessary decline are serious for people with chronic or progressive conditions.

Physiotherapy was sometimes provided at school and terminated after the young person with JNCL left school:

Physiotherapy and occupational therapy were stopped when our daughter left the educational system. Today she is 22 years old.

Moreover, the provision of physiotherapy seems to be related to how the effects are measured. Relating the provision of physiotherapy to observed developmental gains is not appropriate for individuals with JNCL. It is often difficult to measure effects of proactive and precautionary measures. The provision of physiotherapy and extra physical training may rather be related to aims of maintaining the physical condition, such as delaying or reducing ongoing decline or supporting engagement in activities in daily life. It may also be related to support of the general health and physical and mental well-being of the person. It is therefore unfortunate that these interventions are terminated when the individual is not showing the expected progress. Delaying a forthcoming deterioration is outside the traditional aim of interventions for children. There are thus urgent needs to build insights and knowledge about JNCL and similar diseases in organizations dealing with such services.

The positive effects of participating in physical activity were evident not only from the parents' evaluations in the present study, but also from the participants' own interest in such activities. The results indicate that physical activities were a main interest for most of the younger participants, an interest that sometimes

lasted into adulthood. Ninety-six percent of the seven-year-olds were interested in physical activities and 51 percent of the 16-year-olds. Moreover, thirty-two percent had physical activities as a main interest at 22 years (note the difference between having an interest and actual doing the activity of interest). The interest seemed to decline over age for individuals with JNCL but this is also true for the general population (Sallis, Prochaska, & Taylor, 2000). Many persons without disabilities become less interested in physical activities with age and physical activities are replaced by other interests. However, in spite of the considerable difficulties related to their developmental decline, some of the participants in the present study maintained their interest in physical activities. The following quotation from a parent of a child with JNCL illustrates that participation in physical activities may have many perspectives:

Being involved in physical activities provides my son with a sense of mastery, pride and having a meaningful life.

In the present project, the most common interests involving gross motor activity for individuals with JNCL were riding, swimming, dancing, football and outdoor life. An active physical lifestyle may promote social interaction when such activities are shared with others.

Some factors related to physical activity in individuals with JNCL

Physical activities may be organized, such as doing sports or physical education, or attending a gym club, or non-organized, such as playing with friends, walking to school or climbing trees. The total amount of organized and non-organized physical activities is an index of the individual's level of physical activity (Van Duyn, McCrae, Wingrove, Henderson, Boyd, & Kagawa-Singer, 2007).

The health benefits of physical activities depend on the duration (the amount of time spent in one activity session), the intensity (rate of energy expenditure per time unit) and the frequency (the number of exercises per time unit) of the activity. The physical status is measured in different ways, including muscular strength, endurance and oxygen uptake. The endurance is dependent on the heart, lungs, blood flow and blood vessels (Tremblay, Colley, Saunders, Healy, & Owen, 2010).

The flexibility of movements and range of motion of joints also influence the physical status (Jeremiassen, 2016). The joint's flexibility or motion range is normally good in children but tends to decrease with age or when particular

movements are not repeated frequently. The length of a muscle and tendons restricts the range of a motion, as stiffness in joints caused by other reasons. Severe forms of stiffness – joint contractures – may cause great pain and immobility. For persons with physical disabilities, it is particularly important to maintain flexibility in joints that are seldom used.

Many countries offer recommendations for physical activity and it is usually recommended that 5- to 17-year-olds should accumulate at least 60 minutes of moderate-to-vigorous physical activity daily (Janssen & LeBlanc, 2010). More than 60 minutes of physical activity may provide additional health benefits. The World Health Organization (2010) recommends that most of the daily physical activity should be aerobic exercises that make one perspire and breathe harder, which elevate the heart rate, resulting in quicker and more efficient delivery of oxygen throughout the body. Aerobic exercises include walking, swimming, running, skiing or dancing for at least ten minutes at a time. Maintaining a physically healthy lifestyle requires paying attention to the duration, intensity and frequency of exercise activities. The overall principle behind physical fitness is simple: the body becomes a mirror of the lifestyle and motor capacity can only be built through motor activity.

Motivation and interest for participating in physical activities are often established at an early age and may have long-lasting effects. Education and in particular physical education may contribute to establishing such interests in young children, but it is important that the activities are associated with excitement and having fun (Wuest & Bucher, 1999). The results presented above show that individuals with JNCL can sustain interest and engagement in physical activities, even when impairments become more severe. It is recommended that the school, in close collaboration with parents, takes the initiative as early as possible to promote interest in physical activities in children and young people with JNCL. Many exciting activities are accessible even after the onset of a severe visual impairment. Judo, swimming, goal ball, outdoor activities, gymnastics, wrestling, dancing, cross country skiing, tandem biking are activities that do not require vision or much orientation (see chapter 16).

Physical activities for individuals with JNCL

Motor skills vary among individuals with JNCL. Some of the motor skills learned and automatized in early life are maintained past adolescence and into adulthood. Some persons with JNCL have maintained motor skills by practicing advanced activities, such as downhill skiing, skating, fishing and hunting into emergent adulthood, despite their visual, motor and cognitive impairments. Maintaining



Participation in active outdoor life may continue into adult age.

participation in such activities may depend on personal motivation but also on sufficient adaptation and support (see Chapters 16 and 23). To create an early foundation for an active physical lifestyle is a core issue in habilitation for children with JNCL.

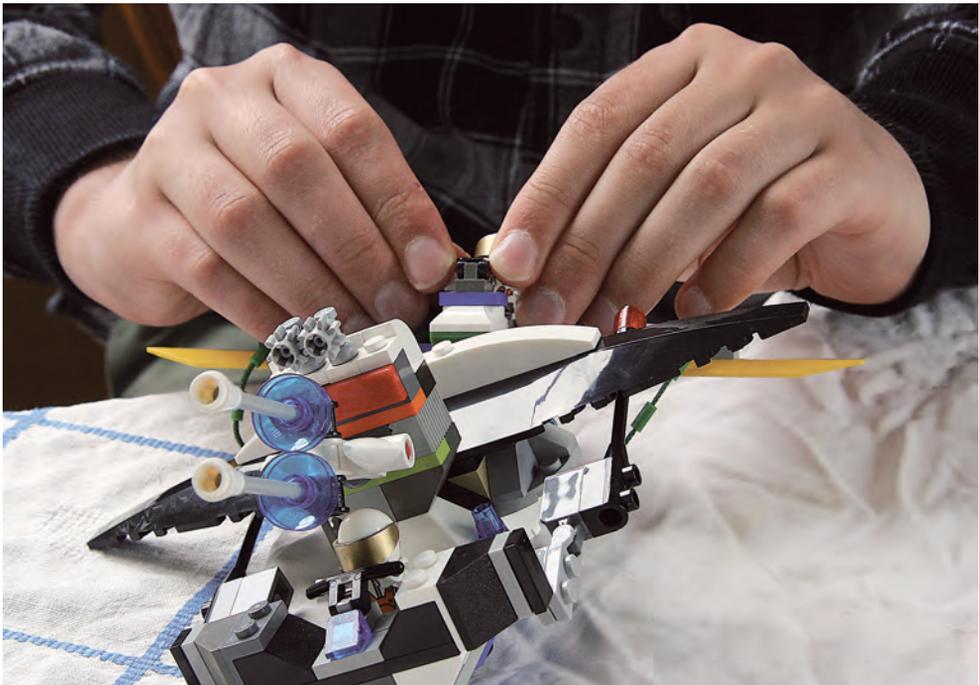
Children and young people with JNCL may need support to develop and maintain fine and gross motor skills and to develop a healthy lifestyle. This area should be a core element in the adapted or special curriculum and the Individual Educational Plan (see Chapter 11). The Educational Development Observation Tool (see Chapter 10) includes observations of motor performance in various situations. The results of the assessment may indicate a need for a physical activity bank where activities are listed and timetabled (see Chapter 23). It is an overall aim that the activities should take place in social and inclusive settings, at school and at home or in the residence. Early motor interventions may have positive life-long consequences for the individual (Jeremiassen, 2016; Wuest & Bucher, 1999).

An overview of physical activities that children and young people with typical development are involved in, in terms of content, extent and frequency, may constitute a basis for planning of physical activities for individuals with JNCL. Such an overview might show that peers usually participate in soccer training twice a week, physical education at school three times a week and scout activities

every Saturday. They might walk or bike to school on a daily basis, visit the playground every second day, go shopping once a week and help the family with gardening every weekend. Such activities constitute the physical lifestyle or baseline for children in general and they contribute to the children's motor development. Not all of these activities are suited for children with JNCL, but they represent a way to explore activities that may be selected for further consideration. For instance, if children with a typical developmental course walk or run 50 kilometers per week and same-



Exploring objects with the fingers.



Constructing with Lego.

age children with JNCL move five kilometers per week, this may indicate a need for intervention. The concern is to compensate for loss of activities caused by the impairments and to provide support and activities. This will require adaptation as well as organizational measures, for example extra hours in physical education or support to enable active participation in the local swimming, judo or riding club. Walking to school or doing chores (e.g., fetching the milk or disposing of the garbage) and other routine activities at school and home will usually require planning and organization.

Fine motor skills are important for children with JNCL. For example, such skills are needed in reading and writing braille or Moon, exploring objects and tying the shoelaces. The hands and fingers become the individual's "eyes" when vision is lost. Examples of fine motor activities are construction play with Lego, doll play, playing cards (braille marked when needed), and everyday activities like cooking and dressing.

Playing the piano or other instruments are also good exercises for developing fine motor skills. Shooting may coordinate fine motor skills and sound location. Children and young people with JNCL may engage in such activities at a frequent and regular basis (Jeremiassen, 2016).



Shooting with the help of audio signals.

Movement restrictions and stretching

Stretching means that muscles are deliberately flexed or stretched in order to maintain or improve the range of motion. It can be performed individually or together with a partner. Stretching will increase the elasticity of the joints and improve the muscle tone, which may help maintain posture and reduce motor decline (O'Sullivan & Portney, 2014). The introduction of stretching exercises is recommended for young children with JNCL to avoid slow developing or non-observable decline in range of motion. Stretching exercises for hips, knees, wrist and shoulders should be prioritized.

Jeremiassen (2016) emphasizes the need for regular assessment of joints and muscles from an early age in children with JNCL. The main aim of the assessment is to register possible ongoing restrictions and limitations in movements. She describes three levels of movement restrictions: (1) full movements (no restrictions), (2) moderate movement restrictions and (3) severe movement restrictions. Restrictions can be caused by motor impairment but may also result from a sedentary lifestyle where joints and muscles are not used on a frequent basis. Most movements restrictions are a result of both, and there can be evidence of a downward spiral where a lower activity level contributes to decreased functionality, which in turn further limits activity. Planning is essential to avoid or reduce this spiral.

Inactivity or inability to use certain movements and joints may result in shortened muscle fibers, increased reflex activities and spasticity of the muscles involved. A severe condition may inflict pain and have other unpleasant consequences for the individual. It is important to be vigilant when evaluating hip and knee movements, since restrictions on these joints may result in problems with walking, caused by an inward rotation of legs or other joint anomalies. Beginning at early age, there should be regular examinations for the following joint functions: (a) elbows: degree of flexion, tension, and inward/outward rotation; (b) wrists: degree of flexion and tension; (c) fingers: extensions; (d) shoulders: ability to elevate and rotate arm; (e) knees: flexion and tension; (f) hips: stretching and inward/outward hip rotation; and (g) ankles: flexion and stretching the ankles. Movement restrictions on Level 2 (moderate movement restriction) and Level 3 (severe movement restriction) should be met by appropriate measures to maintain or improve the flexibility in joints and muscles. Measures may include exercises requiring active and passive movements. Difficulty achieving a full range of motion can be observed at a relatively early stage of the JNCL disease, especially in weight-bearing joints, such as hips, knees and ankles. Activities that will preserve optimal range of motion and mobility are therefore important in early stages of the disease, including activities requiring passive movements, and participation in all-round activities that require all the muscle power available.

The problem with balance and body posture is partly caused by restrictions in the joints and muscles (Jeremiassen, 2016). The joints become stiffer with aging and it may only be possible to execute movements at a slower pace. Preventive interventions should be introduced to avoid an accelerated development of stiffened joints and movements and maintain the best possible posture. Severe forms of stiffened joints and muscles will entail difficulties in undertaking certain body positions, such as stretching out when resting, often combined with pain and discomfort for the individual. A physiotherapist or JNCL counselor should be contacted when such problems are observed. In the present project (Appendix A), several parents and professionals commented that participants with JNCL had been experiencing pain when they went to bed because they were unable to stretch their legs in a lying position. Such problems can to a large extent be avoided with appropriate interventions.

It is important to observe the joints and muscles that are used for posture, walking and movements of the arms and hands. Joints and muscles in the neck, back, hip and knees are important for maintaining posture and walking. The joints and muscles in the arms, hands and fingers are important for daily activities like dressing, reading braille or playing games. Any signs of limitations in these joints and muscles should therefore be met with interventions. The joints should be flexed and stretched and the muscles strengthened or maintained on a frequent basis.

Active movements

Active movements are executed by the person and may have positive effects on the exercised joints and muscles and contribute to maintaining joint mobility and muscle strength. Exercises with active movements can be a part of a training program and executed independently or with support from helpers. The exercises will not improve the movement range or prolong the muscles or tendons, but they can be used to maintain the current movement range.

Active movement exercises can be organized as ordinary body-building programs in a gym or in the classroom or living room. Both the exercises and the results should be registered, documenting both positive changes and challenges. Records that show improvement may increase motivation both in the person and the involved staff. However, a declining curve should never be used as a reason for stopping the exercises, but instead for adapting them. Decline is inherent in the disease, so the aim of the exercises is to slow down the decline as much as possible.



Active movements.

Passive movements

Some movements may be difficult to execute for individuals with JNCL, particularly in the later stages of the disease. Contractures will result from a failure to exercise joints and the corresponding muscles (see Chapter 7). The joints may become immovable if not met by appropriate preventative measures, including a regime of passive range of motion exercises (Jeremiassen, 2016). Passive range of movements implies that a helper is performing the movements of the joints and muscles while the person remains passive. Passive movements can contribute to maintaining and extending the movement range in a joint and are especially important when a joint has become stiffened due to inactivity, such as when a person with JNCL is unable to exercise the joint and muscles. However, passive movements should be made by trained professionals or appropriately trained family members or others, since they may entail pain and discomfort.

Exercises with passive motions can be used with most joints (e.g., the wrist, elbow, ankle, fingers or hip). They should be performed in pleasant settings, such as when the person is listening to music or an audio book, and should be organized as a routine, for example twice a day or as needed. The aim of passive exercises is to prevent both the restrictions on movements or improve mobility, and the pain



Passive movements of the hip.

that may be caused by an immovable joint. The exercises can be conducted by teachers, parents or assistants with guidance from a physiotherapist or another competent professional. The program should be described in writing and if possible with illustrations, and should also include how long time each exercise should take.

Exercising cardiovascular endurance

The motor impairments related to JNCL may make aerobic training difficult when the person is no longer able to walk or stand. However, aerobic capacity is of great importance and should never be forgotten when planning interventions for individuals with JNCL. The functions of the arms and hands are usually maintained longer than functions supporting walking, posture and balance (see Chapter 7). Exercising the upper body may therefore replace activities requiring the aforementioned skills. For example, a rowing machine is found in most gyms and allows exercise of the upper body without making demands on balance, and the motion resistance can be adapted to the individual's capacity. There are sledges with skies or wheels, and they can be used both in winter and summer. The sledge does not make demands on balance and is moved forward with arms and poles (different poles for winter and summer).

Driving a manual wheelchair may also provide aerobic training. There are special wheelchairs for aerobic exercises, which are lighter than ordinary wheelchairs and can be adapted to individual needs. The flow rate or load in these exercises can be increased by practicing uphill or on soft ground.

Exercises in the water are also a good way of promoting cardiovascular endurance when balance problems make other exercises difficult. The water provides buoyancy and support for balance but the resistance in water also makes exercises and motions a bit harder to perform, thereby making the cardiovascular endurance training more effective. Some parents and staff in the present project commented about participants with JNCL who were able to swim after their ability to walk had been reduced or lost, indicating that exercises in the water may be a good option for them. It is recommended that aerobic training is scheduled four to five times per week, or even more often when possible.

Two stories

This part presents two stories about activities and physical education and intervention. The first is told by a father, and the second by a physiotherapist who started to offer physiotherapy to a young man with JNCL aged 20 years.

The father: Keeping in shape and having fun

This is a story about "Harry", a young man of 28 years. Harry was always active and became interested in sports when he was quite young. For instance, snowboarding was learned at a time when he still could see. His body never forgot it and he was able to practice snowboarding until he was 27 years old. One of his helpers at a winter camp recently related that he met Harry for the first time when he was 12 years old. At that time he was able to snowboard downhill on quite demanding trails a number of times in a row together with an assistant and with a slalom pole between them for steering purposes. At the age of 27, Harry could move five meters with the snowboard with the help of three assistants. Still, it was very clear to a proud young man that he had been snowboarding that day.

In addition to snowboarding, Harry was also a proficient cross-country skier. As long as the tracks are well made and clear he could continue to ski for almost as long as walking was possible.

Harry was also able to follow a normal judo curriculum for seven or eight years but had to give up this sport when he was around 20 years old. Before finishing his judo career, he was awarded a blue belt. Judo is the preferred martial sport for many people who are blind, because body contact to a large extent compensates for the lack of vision.

Swimming has always been a favorite of Harry's, and he is still swimming today. In his younger days, he agreed to work towards a specific goal and he would be awarded a merit badge if he was able to meet the ordinary requirements for obtaining the badge. He had to do 1200 meters breaststroke and 600 meters on his back, float 300 meters, dive to the depth of four meters, swim under water 15 meters, undress in the water and show proficiency in life saving techniques. In addition, he was required to dive from a height of three meters (the toughest challenge). Through enthusiasm and stamina, Harry was able to accomplish all of this over a period of two years. A proud young man at the end of the day!

Wall climbing is another sport that over the years proved to be a good activity for Harry. He participated in this sport on a regular basis until he was 26 years old, and still occasionally climbs when he has an opportunity. Wall climbing can be adapted to the individual, made hard or comparatively easy. Getting to the top of a wall and ringing the bell is satisfying, also for those who watch!

Many individuals with JNCL enjoy horse riding. Harry started riding at the age of eight or nine and still rides once a week. Balance has become a challenge, and his saddle has been modified to accommodate balancing support. Horse riding should now avoid going downhill and he uses the same horse always. Harry and his horse make a fine team!

Finally – Harry enjoys a number of other physical activities. At this stage in his life he is dependent on suitable equipment and good helpers. To assist him in his endeavors, he has a tandem bike with a small motor to help climb the steepest hills. He also has a toboggan or sledge which can be modified to accommodate skis, skates and wheels.

The physiotherapist: Growing with training

The young man was 25 years old when this text was written. He lived in a small rural community offering a lot of activities and physical training. He was involved in activities like hunting, fishing and sports, and participated in parties and fun. He had a good network of friends sharing his interests since he was a young boy. Today, he has his own "company" that is cutting, packing and selling firewood. The company is supported by his parents with participation of friends from the neighborhood. The small rural community has always supported the young man's schooling in the best possible way, and he had excellent teachers from the early school years on.

The young man received the JNCL diagnosis when he was seven years old and he became blind in a very short period. The collaboration between the young man, his teachers and the family always worked well. Today he is living in a staffed apartment. He has had a "normal" life where he has been exposed to similar challenges as his peers in a safe environment, supported by friends, engaged staff and family.

Physical activities and a healthy lifestyle were given priority in the young man's life from the very beginning. However, he found physical exercises rather boring after he became an adult. Sessions of planned physical activities were often dropped in favor of more pleasurable sedentary activities after he left school.

The role of the consulting physiotherapist (here: counselor) was to guide the local staff and a local physiotherapist. Plans were made to change the young man's sedentary lifestyle in close collaboration with himself, the local staff and his family. The overall aim was to improve his physical status and specify appropriate exercises. This process started up with a local workshop in collaboration with the project. Different themes were elaborated and reviewed in this workshop and physical activity and training were given priority together with a few other themes. The staff was taught about the importance of physical fitness for individuals with JNCL, what activities to focus on and how physical training could be integrated into the young man's life.

It was decided that physical training should be a part of the young man's working day. Physical training should also be an important life content during afternoons and weekends. Training programs and corresponding schedules were specified:

- *Hard training three times per week during working hours in a gym close to the young man's workplace.*
- *At least 30 minutes of physical activity every day beyond working hours with low to middle pace of aerobic activities, in accordance with recommendations from the health authorities.*

There were follow-up meetings with the counselor, the staff and the physiotherapist every six months. A tool for fitness review and evaluation was developed, which included measures of muscle strength and movement ratio in joints. The results were used to adapt the young man's exercises to his current situation. New baselines were set every sixth month on the basis of an evaluation of his walking ability.

There was little or no research to be found on JNCL and physical exercise. The counselor borrowed ideas from studies of epilepsy and exercise, dementia and exercise, and from her own studies on cardiovascular exercise (endurance training) and its effect on muscle strength and movability of joints. It is a common myth that physical activity may trigger epilepsy (Brna, Gordon, Woolridge, Dooley, & Wood, 2017) but research shows the opposite: physical activity may actually function as an anti-seizure measure (Häfele, Freitas, da Silva, & Rombaldi, 2017; Pimentel, Tojal, & Morgado, 2015). Research also shows that physical activities may have positive preventive effects on cognition and well-being in elderly people with dementia (see Chapter 5). However, the health effects of physical fitness training are dependent on frequency (how often there are sessions), duration (time length of the sessions) and intensity

(how hard the sessions are). In other words, training has to be carried out frequently, have a minimum duration per day and be executed with some intensity.

Good routines do not make themselves

Good routines do not make themselves; they must be implemented over time. It is a continuous process where needs are addressed by new staff members and in accordance with the progress of the disease. A training card was developed where the young man, the staff, the family and the counselor could monitor the exercises and the physical status of the young man. The training card became a highly motivating tool for all involved partners. It described his current physical activities, the number of series to be performed, the weight or load to be used in different exercises, and so forth.

Physical training became a good routine in the young man's life. He attended all the training sessions with joy. From having lukewarm feelings for physical exercise, he became very devoted to physical training. Physical exercise became a natural part of his life and today he is in a better physical shape than at the beginning of the program five years ago.

The importance of engagement

The local physiotherapist had over time acquired experience and knowledge about the diagnosis. He was very engaged and his role was to support staff, inspire them, and be available when needed. He was impressed with the staff and reported that training took place in an inclusive setting where the staff was not acting as instructors but as training fellows. The staff and the young man are doing similar activities and this has had a positive effect on the young man's motivation.

Why all this training?

The main aim of training is to delay the emergence of motor problems caused by the disease, such as reduced mobility, forward bending posture, arthritis (with joint stiffness and pain) and shortened muscles. The person-centered question was how the future challenges of the young man could be met in the best possible way. One important goal was to develop and maintain walking. The training built on the assumption that strong muscles, good flexibility in joints and best possible fitness would create reserves to delay future declines in motor skills.

The tool for fitness review and evaluation was used to measure the young man's muscle strength and search for possible problems with arthritis and shortened muscles. A six-minute walking test was included to measure the aerobic capacity. Altogether, the information collected with the instrument provided a comprehensive picture of the young man's physical fitness and possible changes from last measurement, and constituted a basis for evaluating the need for adaptations of existing exercises or for new exercises.

Achievements

The main aims of the physical training were increased aerobic fitness to support the young man's working capacity to meet daily demands and strengthened muscles and flexibility in the joints to support his walking and prevent falls.

- *Aerobic training was performed three times per week with three sessions each day.*
- *Session 1: 4 x 3 minutes running on a treadmill (4 series of 3 minutes) with a two-minute active break (resting by walking). Speed 7 km/h on a treadmill with a climb of six percent.*
- *Session 2: 2 x 6 minutes running on a treadmill with two minutes active break (resting by walking). Speed 6.5–7 km/h on a treadmill with a climb of four percent.*
- *Session 3: 10 minutes running on a treadmill. Speed 11 km/h on a treadmill with a climb of one percent.*

The young man's speed (frequency of steps) and distance have increased during the last eight-month period. For instance, the speed has increased from six to seven km/h in Session 1, and the distance has increased by 50 meters per interval. The young man is therefore able to run 200 meters longer than before in one session with four intervals. The running speed was 10 km/h in Session 3; today the running speed is 11 km/h. The walking test also confirms an improvement in fitness. Today, the young man is able to walk 820 meters in six minutes, compared to 580 meters eight months ago – in spite of the inherent declines of the disease.

The main focus of the muscle training has been quality rather than weight. Too heavy weights would have had a negative influence on the quality of performance. The quality of all exercises has increased and today the young man is working with heavier weights than before.

The young man had an increased inward rotation of the legs, feet and hips when sitting, standing and walking, which is a common problem in individuals with JNCL. This was a possible sign of weakening muscles, which was met by exercising the outward rotating muscles. The young man started with a 20 kilo load, today he is able to work with 35 kilos, and he has progressed from three times ten repetitions to six times ten repetitions. The quality and coordination of the performance involving both legs simultaneously is also a lot better today. Measures of joints and muscles show an improvement on the left side over the last eight months, and also a stabilization of the decline in the right side.

Full stretch and movements in hip, knees and ankles are important for maintaining walking. The young man has bending tendencies in the knee and hip and has therefore made exercises to strengthen the muscles attached to the knees and hips.

In the beginning he was not able to do three times five repetitions with a ten-kilo load when exercising the hip stretching muscles. Today, he does three times ten repetitions with a ten-kilo load and executes the exercises with better quality. Similarly, he was not able to do three times ten repetitions with a ten-kilo load when exercising knee stretching muscles. Today, he is doing three times ten repetitions with a 15-kilo load with better quality. It took only two to three months of training before improvements became noticeable. Running on the treadmill and walking tours in afternoons and weekends were supporting factors on this positive development. The young man's posture has improved as a result of these activities, in combination with exercises for strengthening the muscles in neck, back, shoulders and arms.

Physical exercises have become routine and are very positive activities for the young man. He is very proud of his improvements and of being strong. He has a very competitive attitude, he likes to use heavy loads and test his limits. Today, the young man is requesting training on weekends when he is normally free from training. Physical training has become a meaningful, inspiring and social activity for him.

References

- Brna, P. M., Gordon, K. E., Woolridge, E., Dooley, J. M., & Wood, E. (2017). Perceived need for restrictions on activity for children with epilepsy. *Epilepsy and Behavior*, 73, 236–239.
- Castro, S., & Palikara, O. (Eds.) (2018). *An emerging approach for education and care: Implementing a worldwide classification of functioning and disability*. Abingdon, UK: Routledge.
- Donnelly, J. E., Hillman, C. H., Etnier, J. L., Lee, S., Tomporowski, P., Lambourne, K., & Szabo-Reed, A. N. (2016). Physical activity, fitness, cognitive function, and academic achievement in children: A systematic review. *Medicine and Science in Sports and Exercise*, 48, 1197–1222.
- Häfele, C. A., Freitas, M. P., da Silva, M. C., & Rombaldi, A. J. (2017). Are physical activity levels associated with better health outcomes in people with epilepsy? *Epilepsy and Behavior*, 72, 28–34.
- Janssen, I., & LeBlanc, A. G. (2010). Systematic review of the health benefits of physical activity and fitness in school-aged children and youth. *International Journal of Behavioral Nutrition and Physical Activity*, 7, 40.
- Jeremiassen, R. E. H. (2016). *JNCL og fysisk aktivitet (JNCL and physical activity)*. Trondheim, Norway: Statped Midt.
- O'Sullivan, S. B., & Portney, L. G. (2014). Examination of motor function: Motor control and motor learning. In S. B. O'Sullivan, T. J. Schmitz & G. D. Fulk (Eds.), *Physical rehabilitation, Sixth edition* (pp. 161–204). Philadelphia, PA: F. A. Davis.
- Pimentel, J., Tojal, R., & Morgado, J. (2015). Epilepsy and physical exercise. *Seizure*, 25, 87–94.
- Sallis, J. F., Prochaska, J. J., & Taylor, W. C. (2000). A review of correlates of physical activity of children and adolescents. *Medicine and Science in Sports and Exercise*, 32, 963–975.
- Tremblay, M. S., Colley, R. C., Saunders, T. J., Healy, G. N., & Owen, N. (2010). Physiological and health implications of a sedentary lifestyle. *Applied Physiology, Nutrition and Metabolism*, 35, 725–740.

- Van Duyn, M. A. S., McCrae, T., Wingrove, B. K., Henderson, K. M., Boyd, J. K., & Kagawa-Singer, M. (2007). Adapting evidence-based strategies to increase physical activity among African Americans, Hispanics, Hmong, and Native Hawaiians: A social marketing approach. *Preventing Chronic Disease, 4*, 1–11.
- World Health Organization (2010). *Global recommendations on physical activity for health*. Geneva, Switzerland: World Health Organization.
- World Health Organization (2018). *Active: A technical package for increasing physical activity*. Geneva, Switzerland: World Health Organization.
- Wuest, D. A., & Bucher, C. A. (1999). Historical foundations of physical education and sport. In D. A. Wuest & C. A. Bucher (Eds.), *Foundations of physical education and sport, 13th edition* (pp. 146–193). Boston, MA: WCB McGraw Hill.

16

Coping with Everyday Life

*Bengt Elmerskog, Stephen von Tetzchner,
Marja-Leena Forssas and Tarja Eskonen*

After a child has been diagnosed with JNCL and the illness progresses, people surrounding the child may increasingly do things on the child's behalf because they want to protect the child from potentially dangerous situations or to get things done faster than the child can do unassisted. In addition, they may underestimate the child's self-help skills and other abilities. This may over time lead to excessive dependence on help from adults, potentially resulting in learned helplessness and passivity. An alternative approach is to support autonomy and independence when skills decline. Baltes and Baltes' (1990) model of goal selection, optimization and compensation (see Chapter 2) provides a useful framework for the planning process. Independence, with or without assistance and support, is a basis for self-confidence and self-efficacy in children and adolescents and for coping with daily life (Aro, Siiskonen, Niemelä, Peltonen, Stenroos, & Kulmala, 2007; von Tetzchner & Martinsen, 2000).

The progression of vision loss in JNCL can be rapid and the consequences dramatic. There may be a loss of activities, friends, participation and independence. Blindness will restrict the possibilities of acquiring information from the environment. Vision is holistic and immediate, just a short glance may give an overview of what is happening, of the topography and the on-going activities in the near environment. Vision is a core sense behind independence and participation. Additional declines in cognition, communication or motor performances will indeed make independence and participation more complicated, and it is important to meet these challenges with targeted measures to achieve best possible participation and coping in daily life. Compensation for a visual impairment is a matter of learning new behaviors supplemented by making physical adaptations in the environment. The child must learn how to use touch and hearing in a different way and to accumulate and memorize received information to build up a holistic understanding of the environment to facilitate independence and interdependence (see below). Further, compensation is also about making participation in meaningful activities available for persons with JNCL.

Physical adaptation for people with severe visual impairment is about making features of the environment available through hearing and touch. For instance, a green area on the wall is not available through touch or hearing but giving this area a tactile texture will make the area perceptible for persons with blindness. A door that is not noticeable from a distance for a person who is blind, can be rendered perceptible with the addition of an audible signal.

Increasing students' determination, autonomy and independence in coping with life towards adulthood is an overall aim of the school, implying that young people will become able to accomplish daily tasks without help from others. These are indeed important goals in education, but goals of independence will sooner or later entail challenges and difficulties for individuals with JNCL. However, coping with everyday life has a wider perspective than independence, and this is of special significance for persons with JNCL.

Independence and interdependence

The core of the wider perspective on coping is acting as a *driver in one's own life* and not *being a passenger*. Being a driver is an essential element of self-determination and quality of life. There are many ways to show self-determination, such as when children choose to break up with friends outside of school time, refuse to do what they are told to do, and play on their own. These actions may be viewed as declarations of being a driver in their own life. When children do not want to be a part of a group activity, the reason may be a lack of interest or insufficient skills to participate. Alternatively, they may not enjoy the social setting or just want to demonstrate their self-image or role. Such behaviors may also be a way to try to obtain recognition. Protests and refusals may be perceived as annoying by others but are of importance for the personal development of all children.

Experiencing being in the driver's role in their own life is of special importance for children and young people with JNCL. For them it may be a source of motivation for further learning when they experience that it is possible to act as a driver despite reduced independence. Being a driver in one's own life requires self-governed attention and action, which is also a foundation of being active in learning and development. Doing things that are determined by someone else on your behalf does not require similar processes. For instance, it is more effective to learn the route between two places if one is driving the car, compared with being a passive passenger. The concept of being a driver also includes making one's own choices in life. Choice making is likely to increase attention and mental effort even if physical independence is not possible.

Typical development proceeds from *interdependence*, doing things together, to *independence*, doing things by oneself (Greenfield, 1994). In the theory of Vygotsky (1962), interdependence is a characteristic of the zone of proximal development, and there may be various degrees of independence and interdependence in different domains (von Tetzchner, 2019). When a person's developmental path involves decline, the process may go the other way, from independence to interdependence, and interdependence is a characteristic of the zone of developmental maintenance (see Chapter 2).

Independence and interdependence are two basic forms of active participation and coping. From the perspective of being a driver and coping with everyday life, independence means achieving personal goals *without support*, while interdependence means achieving personal goals *with support*. In interdependence, there is mutual reliance between two or more persons who are dependent on each other to achieve the chosen goals. It may be a transactional chain of interactions, where one person's actions trigger actions in the other person, and so on. An interdependent relationship will sooner or later be necessary for persons with JNCL to maintain the driver role in their lives, albeit one that resembles a learner-driver. For people caring for a person with JNCL, interdependence requires a strong and persistent focus on autonomy where they give support but *enable* the person with JNCL to be the driver. Carers may need support in learning to play their part in this new relationship based on interdependence.

Autonomy may be defined as a person's ability to make his own decisions, the ability to act on his own behalf without interference from others; it is about self-determination, not self-reliance (Entwistle, Carter, Cribb, & McCaffery, 2010). Autonomy is recognized as important for achieving good mental health (Taylor, 2005). Intervention that supports autonomy may enhance overall adjustment and life quality. The possibility to behave and act autonomously may make people feel and perform better, while lack of autonomy may lead to loss of interest and motivation, as well as sickness and depression. Development of autonomy may not always be an explicit goal in special education, but it is an overall goal for persons with special needs and in particular for persons with dementia (Agich, 2003). Supporting and maintaining autonomy in persons with JNCL require dignity, respect, understanding and time from people in the surroundings when collaborating.

Their early typical development implies that children with JNCL like their peers are acting as drivers in their own lives. They interact with their peers on an equal footing, display the same motor skills, and participate in ordinary childhood activities. The way from independence to dependence in developmental decline may involve several phases, involving compensations, help and support, and change of goals (Baltes, 1997). In earlier phases of decline, technical aids and

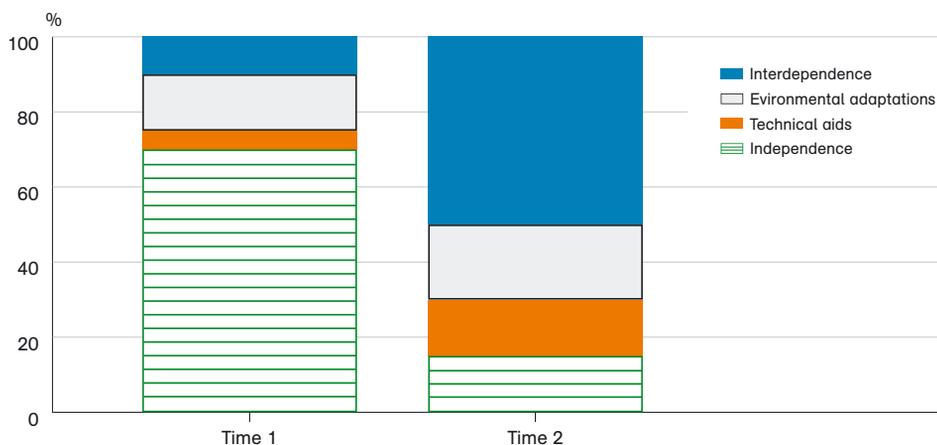


Figure 16.1 Hypothetical development from independence to interdependence, resulting in full participation

environmental adaptation may be enough to maintain everyday functions. Figure 16.1 shows the relative amount of independent functioning, compensation, and help at two points in time. At time 1, the participation of an adolescent boy with JNCL is mainly independent, although with short periods with compensation and help. At time 2, the same adolescent is more affected by the disease. However, he is still participating 100 percent of the time but his participation is dependent on more diverse compensatory measures and especially on collaboration with others through interdependence. Despite the decline, participation is achieved successfully with the help of educational, environmental and other interventions.

In this chapter, the concept of coping is associated with participation in everyday activities. Participation in activities will require the accomplishment of a series of tasks through independence when this is possible and desired, or interdependence when support is required and desired. Allowing for independence and interdependence according to the needs will enhance personal autonomy and coping with everyday life.

Education for coping with everyday life

Orientation, mobility and skills related to activities of daily living are important elements in special needs education for students with visual and cognitive impairments and for achieving independence and interdependence. Goals related to interdependence and autonomy typically become important in education when academic and practical performances are difficult to achieve due to the progressive

visual, cognitive and motor declines in children and young people with JNCL. The use of technical aids is also an important element that contributes to the coping process (see Chapter 19).

Orientation and mobility

Severe visual impairment influences orientation and locomotion. Moving around independently requires cognitive resources and is one of the greatest challenges for persons with severe visual impairment, and orientation and mobility are always core elements in the education of children with severe visual impairment (Martinsen, Tellevik, Elmerskog, & Storliløkken, 2007). Understanding the physical environment is essential for all children. Limitations in orientation will influence independence at home and in unfamiliar areas. Teaching of orientation and mobility is therefore crucial for transitions and the child's understanding and knowledge of the respective environments (McConachie, 2018).

There are different definitions of orientation and mobility. Lowenfeldt (1954) defines orientation as the ability to recognize the surroundings and their temporal and spatial relations, and mobility as self-propelled locomotion from one place to the other. Törrönen and Onnela (1999) define orientation as the ability to determine one's position in an environment based on awareness of the body and environment through use of the remaining senses. They claim that the bases of mobility include perception of body and space, control of posture and movements, balance, and basic skills of moving around. Elmerskog, Martinsen, Storliløkken

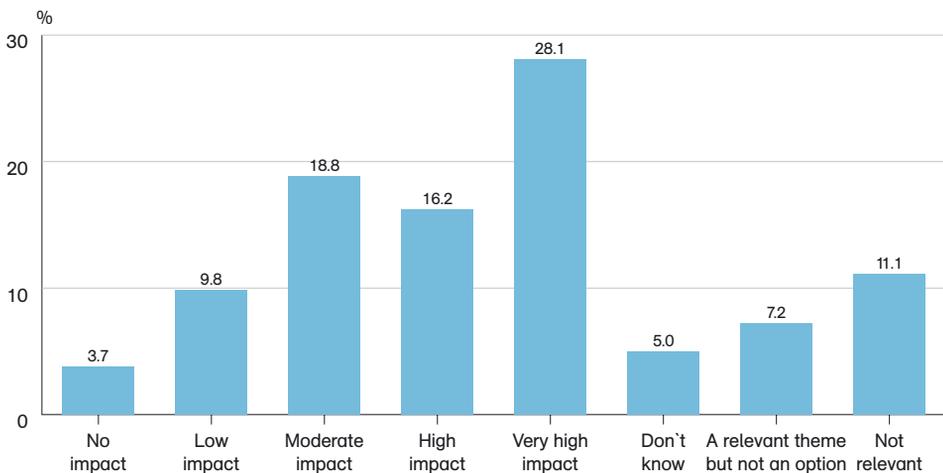


Figure 16.2 Parents' evaluation of impact of orienting and mobility training (percent of answers) (N=109)

and Tellevik (1993) define orientation and mobility as goal-directed activities in relation to objects in space.

Orientation and mobility education may include mobility route learning, use of sighted guides, searching skills and protective techniques, efficient use of the white cane and physical adaptation of the environment.

Parents in the present JNCL project (Appendix A) were asked whether their child had received orientation and mobility training, and if the training had any positive impact on the child's daily life. Of the 109 parents who answered this question, 76.7 percent answered that their child had received this kind of training, 11.1 percent said it had not been relevant, and 7.2 percent had not received training even if it was needed. Five percent said they did not know, but it is not clear if «Don't know» meant that the parents did not know if the child had this form of training or were uncertain if the orientation and mobility training had had any positive impact. Just over 44 percent said that the orientation and mobility training had a high or very high impact on the child's daily life, while 19 percent reported a moderate impact. Nearly 14 percent found that the orientation and mobility training had little or no positive impact on their child. The low impact may indicate that vision was still good or that the training had little effect. The mobility and orientation training was introduced at a mean age of 9.7 years.

There are some technical mobility aids designed for use with children and adults with multiple disabilities (Lancioni, Sigafoos, O'Reilly, & Singh, 2013; Mulloy, Gevarter, Hopkins, Sutherland, & Ramdoss, 2014; Zhou, Parker, Smith, & Griffin-Shirley, 2011), but research on the use of such aids by children and young people with JNCL is lacking.

Activities of daily living

Activities of daily living are generally considered as tasks requiring independence in coping with self-care, home and routine activities that most people do every day without assistance, such as using the toilet, grooming, dressing, feeding and community skills like shopping, home making, and so forth (Giebel, Sutcliffe, & Challis, 2015; Prizer & Zimmerman 2018). A clear definition of activities of daily living training is lacking, but training in such activities is often a core element of special needs education. Trainers and support workers are responsible for adapting the environment to the person's needs. The aim of the training is to facilitate an optimal level of independence in daily life. The level of training and adaptation required depends on the student's needs in coping with daily life. The traditional activities of daily living are in this chapter described as "tasks". Such tasks should be part of a meaningful, real-life activity, be functional and have personal value.

For example, activities like having dinner, playing soccer or visiting friends are meaningful for most people. Each of these activities may comprise several tasks: paying for a ticket is a task within activities like «watching a soccer game» or «travelling on a bus», adding salt is a task in the activity «preparing porridge», and using an electronic communication device with a switch is a task within the activity «socializing with friends.»

Real-life activities will be part of adapted curricula when they are related to the daily life of children and young people with JNCL. It is always a risk that interventions not related to real-life activities reflect wasted effort and time, such as practicing free kicks if the person does not play soccer. A holistic approach emphasizes that training and other interventions should be authentic and have ecological validity for the person (Tellevik & Elmerskog, 2001).

A wider perspective on supporting coping in daily life is about making priorities beyond self-care activities, such as play, work or sports activities. Educational interventions, like training new reading techniques or preparing physical adaptations, are identified by making a task analysis of the selected activities. Intervention related to activities of daily living is common in the education of children and young people with visual impairment. In the present JNCL study, parents were asked a) to state whether their child had received this form of training and (b) to evaluate the impact of the training. Of the 106 parents who answered (b), nearly half answered positively (Figure 16.3). However, almost 20 percent had not received training even if they thought it was needed, while 25 percent said it had not been relevant. Nearly eight percent said they did not know,

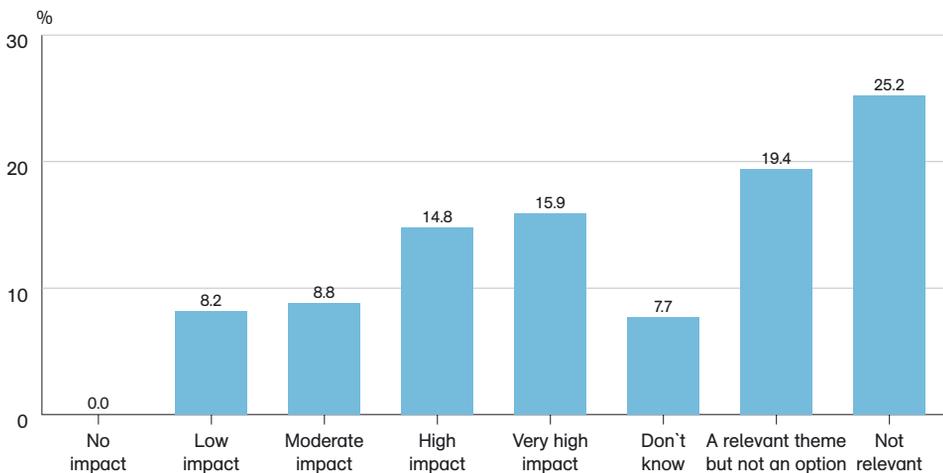


Figure 16.3 Parents' evaluation of impact of training activities of daily life (percent of answers) (N=106)

but it is not clear if «Don't know» meant that the parents did not know if the child had received this form of training or were uncertain if the training had had any impact on their child. Among those who reported an impact of this training (47,7%), 64,4 percent said that the training had a high or very high impact on the child, while about 18 percent reported a moderate impact. About 17 percent reported a low impact on the child from training related to activities of daily living. The mean age for the first training in activities of daily living was 11.1 years.

Training for orientation and mobility or activities of daily living has generally little place in mainstream education but constitutes a central part of special needs education for children and young people with visual impairment. A comparison of Figures 16.2 and 16.3 indicates that the parents considered mobility and orientation more important than activities of daily living. One reason may be that orientation and mobility training is offered to most children with severe visual impairment, independent of the student's cognitive level, whereas training in activities of daily living usually is offered to students with intellectual disability. For instance, in Norway, training in orientation and mobility is a legal right for all people with severe visual impairment, while training in activities of daily living lacks a similar legal status. Training of activities of daily living is usually part of the curriculum for students who are unable to follow the mainstream curriculum. With emerging dementia, activities of daily living should be an integrated part of the education for children and young people with JNCL to maintain independent or interdependent skills.

Decline in independence

The developmental trajectories of children with JNCL differ from those of children with congenital visual impairment or other disabilities. In the early years, children with JNCL are accustomed to managing tasks in their daily life in the same ways as their peers. For instance, the results of the present study indicate that very few children with JNCL have problems with passivity or learned helplessness in early childhood. Despite gradually emerging declines, children with JNCL struggle to be active and maintain the activities of ordinary daily life. Parents and teachers should try to avoid providing care and help when these are not needed, including making unnecessary priorities and life choices on behalf of the child.

The cognitive, visual and motor decline will sooner or later make independence difficult to maintain for individuals with JNCL. Most outdoor and many indoor environments need physical adaptation to make independence or interdependence possible. Physical adaptations include creating topographic patterns like handrails and curbs when needed or marking personal objects in order to make them

recognizable by touch or hearing. Adaptations should be useful even in challenging conditions, such as when it is snowy, rainy, or noisy. Training should focus on goals that can be achieved through interdependence with others. This should start early in life for students with JNCL, because they need to learn appropriate strategies and becoming used to seeking help from people in the surroundings to achieve their personal goals. This is a basic requirement behind autonomy, but it requires a close and persistent collaboration with available helpers, including paid support staff, educators, family, and peers. Coping with everyday life will simply not be possible without such support in later phases of the JNCL disease.

Early intervention related to low vision

The length of the period from the first signs of visual loss to blindness varies, from one year up to ten years according to findings in the present study (see Chapter 4). The remaining vision will influence learning and coping with everyday situations, and it is of great importance that needs associated with level of vision are met by appropriate measures. Many complex tasks are easier to learn when the child has some vision. The child can learn by observing others' behavior when performing tasks, for instance how to tie the shoes or soap the hair, and not least, different kinds of social codes. It is therefore important to try to establish and automatize certain complex skills before the onset of blindness, when this is possible.

The period of decreasing vision will entail several practical problems; the child will need more and more time to accomplish routine activities. Carers should be careful not to take over tasks that the child can perform, even if it takes the child longer to complete them. Maintaining realistic expectations to the child despite the declines is important, for instance in connection with potentially challenging tasks such as fetching items, moving around, zipping up the jacket, and so forth. However, sometimes trade-offs must be made. Life is not only about performing daily routine tasks, but also about having fun. Too much time used on undressing may interfere with a forthcoming desired activity like swimming or playing. Finding a balance among these competing needs of daily life is essential.

Most students with JNCL are rather self-sufficient in independence in their early period of low vision. Interventions in this period of life include providing sufficient lighting and better organization of objects and activities, eliminating hazards, creating adequate visual contrast between objects and their background, and offering opportunities and technology for magnification (e.g., placing magnifiers strategically where they are going to be used, without needing to carry them around). Creating a person-centered, user-friendly and accessible

environment makes life easier and will strengthen the child's or young person's ability to act as a driver in their own life. The creation of a person-centered and user-friendly environment is often limited to areas frequently used by the child with JNCL. Priorities should be given to classroom, bathrooms, working places, kitchen and/or important pathways used by the child (see Table 16.1).

Table 16.1 Some compensatory interventions for a child with low vision

Proposal (date)	Specific area	Type of intervention needed	Accomplished (date)
31/10-17	Bathroom at home	Additional lightening. Eliminating hazards – always close closets and cupboards	12/11-17
31/10-17	Classroom placing	First row in classroom near teacher's desk	02/12-17
12/11-17	Closet 2 at school	Increase lighting above the mirror	05/12-17
05/12-17	School desk	Gooseneck, swing-arm high-wattage non-glare lamps	10/01-17
10/01-18	Home	Night-lights in bedrooms, bathrooms and hallways to improve safety	26/01-18
10/01-18	Stairway+ entrance at school	Increase lighting above the stairs and antiskid flooring on top and bottom of the stairs to avoid accidents	26/01-18
10/01-18	General at school and home	Organization: Store items near the location where they are used. Put away clothing, cooking utensils, books or CDs immediately after use	Ongoing
26/01-18	Books, CDs and keyboard	Mark books and CDs with color codes, large letters or tactile codes for identification purposes	03/02-18
26/01-18	Keyboard	Mark the keys on the PC keyboard with large letters or tactile dots	03/02-18
26/01-18	Dressing room at school	Children's shoes, packages or other items to be removed from floor to wardrobe	03/02-18
03/02-18	Kitchen and bathroom at school	Install non-skid flooring	10/03-18
03/02-18	Classroom	Install door knobs in good contrast to the door color	10/03-18
03/02-18	General	Contrasts: Place dark-colored items against light background. Example: Place black towels in a white-tiled bathroom	Ongoing
03/02-18	Pathway from school to bus stop	Increase lightening. Hand rail	10/03-18
10/03-18	Playing corner at home	Gooseneck, swing-arm high-wattage non-glare lamps. Magnifying glasses	23/03-18

It is advisable to use appropriate expertise when implementing costly interventions that require knowledge about low vision and low vision technology. However, comments from parents and staff in the present JNCL study (Appendix A) indicate that costly devices in many cases were never used because of both the lack of local competence in how to use the device and the rapid reduction of the child's vision, as illustrated in the following quotations:

It was getting more and more difficult for her to see anything at all. At home she had a screen reader and a monocular, but she never actually used them. I don't know but she somehow refused to use these vision aids, and I doubt the school knew how to make best use of this equipment.

He was encouraged to read using CCTV, there were sensory stories as well, but it was not followed up as needed. Some appropriate technology was used around at the time – his loss of vision declined fast – I'm not sure it had any important function for him. So probably earlier days of technology but, it's different then to now really.

At that time however computers and information technology weren't used at schools yet so I taught her at home to write on a computer.

Then, teachers should have some training in how to use the low vision equipment, and the FBA specialist should be able to put in a plan... difficult times.

The period of low vision can also be used for precautionary interventions, that is, for preparing the child for the forthcoming blindness, such as marking the environment and making the child acquainted with features that can be useful later (see Chapter 12). Adults may choose travelling routes suitable for a child who is blind when moving between places the child is likely to travel later. Outdoor environments may be provided with handrails that later can be used as shorelines. The classroom, bedroom and locker room may be prepared for the child's visual impairment, using tactile marking or placing objects on fixed locations. Interesting texts in braille or Moon letters may prepare the child for later literacy skills, as well as keyboards suitable for tactile writing even if vision-based keyboards are an option in the child's current situation (see Chapter 14). Learning to use technical aids (e.g., a magnifier) can be accomplished through game-like activities that children can enjoy with peers and need not be boring or decontextualized (see Chapter 21). The understanding of the surroundings based on early visual memories may make later coping with daily life easier to maintain after the onset of blindness.

Table 16.2 A step-by-step model to support coping in everyday life

1	Assessment for learning and maintaining activities and making priorities
2	Task analyses of selected activities
3	Task analyses of selected mobility routes
4	Implementation of training and other interventions

Step-by-step support of coping with daily life

Promoting coping in everyday life for children and young people with JNCL includes such things as ensuring participation in desirable activities, providing training in how-to-accomplish tasks associated with those activities, and taking steps to make the physical environment user-friendly for each activity. Barriers and other contextual factors that prevent coping are often found on diverse task levels, such as a lack of physical adaptation and support, problems with transportation, and much more. The ultimate educational goal for individuals with JNCL is that they should be able to participate in desirable activities through independence or interdependence.

Analyzing tasks related to desirable real-life activities may help teachers and service providers identify needs for training, support and physical adaptation. Table 16.2 shows a step-by-step model to support coping in children and young people with JNCL.

Step 1 – Assessment for learning and maintaining activities and making priorities

Mobility routes and activities of daily living should imply participation in meaningful real-life activities. For instance, navigating the route to the activity «play at the playground» is the first and last step or sub-goal for participating in playing. The activity may have more routes, such as the route from the bottom to the top of the slide, or from the slide to the sandpit. The basis for the selection of training mobility routes and activities of daily living is consequently participation in meaningful activities.

Setting priorities for interventions based on real life activities entails a holistic perspective that includes consideration of participation, skills needed, necessary environmental adaptation, support, and other interventions. These factors are analyzed in relation to meaningful contexts. Relating teaching and training to common real-life activities makes it possible to refresh memory and

Table 16.3 Assessment form for activities and making priorities

	Activity	Where	Priority date	Goal	Goal achieved
1	Playing jungle gym	Playground at school		Interdependence	
2	Bathroom activities	At school, ground floor	13/02-18	Independence	05/05-18
3	Bathroom activities	At home	13/02-18	Independence	
4	Eating	Canteen at school		Independence	
5	Arriving at school	Cloakroom at school		Independence	
6	Using smart phone	Anywhere		Independence	
7	Visiting Carl	Home	13/02-18	Interdependence	12/06-18

skills in natural situations and avoid having to schedule training at fixed times that lack a similar degree of contextualization. Supporting learning in real-life activities will in many cases also strengthen the student's motivation (Tellevik & Elmerskog, 2001).

Table 16.3 is an excerpt from an assessment sheet of desired real-life activities of an adolescent with JNCL. This kind of assessment will serve as the basis for prioritizing interventions, as well as for introducing new activities that have not previously been available to the adolescent with JNCL.

Table 16.3 contains seven activities. Three activities were given a priority on February 13th, 2018. Activity number 7 – visiting the friend Carl – is a new activity to improve social interaction. Some goals were defined as independent, others as interdependent, meaning that the activity will require collaboration with a supporter. The goal of activity 2 was achieved on May 5th 2018, and activity 7 on June 12th 2018. Assessing real-life activities as shown in Table 16.3 and prioritizing interventions should be completed as a collaboration between school or institution, parents and – if possible – the person with JNCL.

Steps 2 and 4 – Task analysis of selected activities and implementation of training and other interventions

Most real-life activities consist of several tasks, which often should have a predetermined order. For example, going to the cinema may include «identify a film of interest on Internet», «get money for the ticket», «dress», «walk to the bus stop», «travel by bus», «walk from the bus stop to the cinema», «pay for the ticket», and «find the seat.» All the tasks are parts of the same activity – going to the cinema, but require different skills, actions and help, for instance when searching for information on Internet or walking to the bus stop. It is a chain of

tasks that must be performed by the individual – and when needed – assisted by a helper. If the chain of actions or support of tasks is broken at one place it will result in non-participation.

Interventions are defined on the task level and not on the activity level. In the task analysis, the activity is divided into smaller, more manageable components, where skills, barriers in the environment and need for assistance are assessed separately. For instance, to make the environment easier to manage, the coat is marked for identification purposes and should always hang in the same place. Training could be directed at handling the zip or tying the shoes.

The personal need for participation in real-life activities should always be considered stable for persons with JNCL despite their declines. For instance, the need for participation in social settings does not decline because of speech

Table 16.4 Task analysis of bathroom activities for a person with severe cognitive decline

TASK ANALYSES OF BATHROOM ACTIVITIES AT SCHOOL (GROUND FLOOR)				
	Task description	Physical adaptation	Goal	Achieved (date)
1	Walk from classroom to bathroom	Bathroom door marked with wooden block	Interdependence Free guiding	15/05-18
2	Open door turn light on right hand side		Independence	01/06-18
3	Close and lock the door		Independence	01/06-18
4	Locate bathroom sink on left hand side		Independence	01/06-18
5	Mix water to desired temperature	Hot water tap marked with tactile dot	Independence	01/06-18
6	Push soap dispenser 2 times on left hand side	Soap dispenser placed left hand above the sink	Independence	01/06-18
7	Rub hands together		Independence	12/06-18
8	Put hands under the water and rinse soap off		Independence	12/06-18
9	Turn off the water		Independence	01/06-18
10	Dry hands with towel on right hand side	Towel placed right hand side of sink	Independence	18/06-18
11	Locate door, turn light off		Independence	12/06-18
12	Open door walk out of the bathroom		Independence	01/09-18
13	Walk from bathroom to classroom		Interdependence Free guiding	15/05-18

problems, but participation becomes more difficult and requires new training, adaptation and assistance as the disease progresses (see Chapter 13). Task analyses must be revised to adapt tasks, physical adaptation and help in accordance with the various declines. For instance, a red door that used to be identified by vision may require tactile marking after onset of blindness. The cloakroom should be located close to the classroom and a chair should be provided to make it easier to change clothes when mobility and motor problems become more severe.

The result of a well-designed task analysis will be a structured intervention situation, where the same procedures are repeated. At the same time, the detailed level of task analysis must reflect the person's potential. For instance, a severe decline in cognitive and motor skills requires more details compared to less severe cognitive and motor impairments. Table 16.4 shows a rather detailed task analysis for a student with profound cognitive disability. It includes 13 tasks, each reflecting this child's potential and need for training, help and environmental adaptation. A task analysis of the same activity for another child may consist of fewer or more tasks, and greater or lesser amounts of training, help and adaptation.

The goals of the activity shown in table 16.4 are independence or interdependence. The goals of tasks 1 and 13 are movements to and from the bathroom by using free guiding (see Table 16.10). Free guiding means that the child will follow the guide by listening to the sounds produced by the guide in front of him or her. The goal of the remaining tasks is independence. The analysis revealed that the main problem of task 10 was that the towel had not been given a permanent place. Independence was achieved by giving the towel a fixed place. The table shows that all goals had been achieved by September 1st, 2018.

Steps 3 and 4

– Task analyses of selected mobility routes and implementation of training and other interventions

The ability to move around and get from one place to another is essential for all people. The importance of mobility for locomotion is usually not given much attention until it becomes a problem. However, severe visual, motor or cognitive impairment may reduce the individual's possibility to get around if not met with appropriate measures. This requires access to help or learning how to cope with daily locomotion needs. An inability to move from activity to activity, or between places where activities take place, will restrict the person's possibilities for participation.

Orientation and mobility may include different forms of training, such as how to use the white cane, public transport, tactile maps or master routes, as well as how to be guided by sighted helpers, and use of self-protection techniques

(Elmerskog et al., 1993; Jacobsen, 2013; Wiener, Welsh, & Blasch, 2010). In the following section, the focus is on route training because it is of special importance to children and young people with JNCL.

Children with congenital blindness are often given preparatory orientation and mobility training that will lead to later independence and better skills in orientation. They are taught basic concepts such as right and left, improved body awareness, how to move in different terrains, and gross motor functions while walking. The situation for children with JNCL is different. Early orientation and mobility intervention is more about enabling them to travel as they did before the onset of severe visual impairment, that is, with a stronger focus on the child's *life flow* (Csikszentmihalyi, 1997).

Lacking vision places demands on the person's memory and search behaviors. For instance, observing that a particular table has a flat square area, four corners and four legs is done with a single glance by a person who can see. Making the same observation without vision requires investigation or explorative activities using the hands (Fraiberg, 1977; Tellevik, Storliløkken, Martinsen, & Elmerskog, 2007). Information through touch and movements must be accumulated and memorized to build a cognitive representation of the table. To learn new routes also requires movement, searching behavior and memory. Learning new routes based on non-visual cues requires more anticipated and explorative behaviors and makes higher demands on memory than the use of visual cues. The individual must remember what to do at each point along the route to reach the destination. However, when a route is learned and the child or young person master the navigation, less cognitive effort is required (Tellevik et al., 2007). Teaching children with JNCL to move around independently in familiar environments in relation to important activities prior to onset of total blindness will therefore be a goal of high priority. Enabling children with low vision to act as "drivers" when they move around will support their understanding of the surroundings, which in turn supports maintenance of independence and interdependence when vision has been lost.

Teaching orientation

People with and without vision use different strategies in orientation. Perception based on visual orientation may be described as information from a *field*, while perception based on non-visual orientation to a high degree is based on information from a *line* or *tunnel* (Gibson, 1966). To orientate on a *line* or in a *tunnel* means that most information is proximal and limited to the space that can be physically reached with bodily movements.

According to Gibson (1966), visual information tends to be interceptive (the information comes to the person) while tactile or haptic information requires exploration behaviors. Further, in visual orientation the attention is directed

forward when moving, in non-visual orientation the attention is often directed sideways. For instance, a sighted person will walk, drive and orientate towards features in the environment, while a pedestrian who is blind will walk forward but orientate along features in the environment, such as walls or curbs.

A child with JNCL who is becoming blind must learn new ways of orientation, such as using fences and curbs. In the early phase, this may cause problems in independent travelling. The child may not be attentive to physical features and structures that are required for independent travelling and must learn to search for accessible and relevant information in the environment. The teacher must be aware of such challenges and guide the child to orientate along physical recognizable features and structures. Directing the child's attention towards features and structures along the path of travel can be made by knocking on the wall or carefully leading the child's hand to walls, fences or objects along the path. The main aim of teaching routes is to guide or direct the child's attention to predefined features along the route when moving forward.

There are two basic types of orientation in route training: *egocentric* and *topographic* (Ferrara & Landau, 2015; Newcombe, Uttal, & Sauter, 2013). Figure 16.4 illustrates the difference between egocentric and topographic orientation. In topographic orientation the person uses objects or features in the environment as cues to determine her own position. The experience of walking on the left side of a river or using a roadmap for orientating are examples of topographic orientation. In egocentric orienteering the person uses her own body as reference point for orientating and determining the position of objects or structures in the environment. People with vision can easily choose to use both egocentric or topographic references in orientation since vision will provide information that one is a part of a stable environment. The situation is different for people with blindness.

Orientation for persons who are blind should be based on egocentric orientation when learning new mobility routes. The

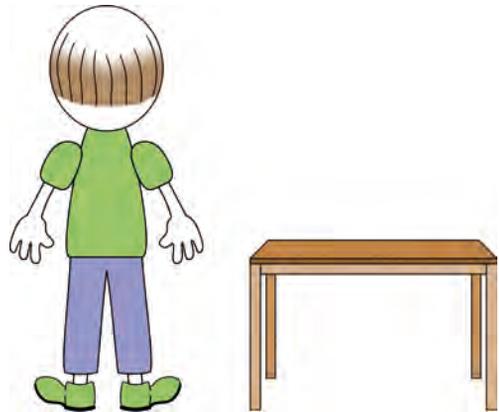


Figure 16.4 Orientation based on egocentric and topographic orientation

The person is observed from behind.
 Topographic orientation: *You are standing on the left-hand side of the table.*
 Egocentric orientation: *The table is on your right-hand side.*

traveler's body should always be used as the reference point in relation to objects, features and structures along the path. Topographic references may require complex cognitive calculation as shown in the following statement: *I am told that I am standing on the left-hand side of the table. This means that the table is on my right-hand side.* Mixing egocentric and topographic orientation in instructions – which is common among people who are sighted – may make route learning very difficult for persons with JNCL. This example also highlights the complexity of language implied in instruction.

Routes

It is a well-known saying that all roads lead to Rome. People who are sighted may use any road leading to their destination. Learning a mobility route means to learn to master one of these roads. A traveler with JNCL will in most cases keep to the route that she or he has learned when not assisted by a sighted helper.

A mobility route is a pre-defined path from a specific starting point to a specific destination. It can be the long road from home to school, or the short path from the kitchen table to the refrigerator. Learning orientation is learning goal-directed attention towards important features in the surroundings to reach a desired location and activity. The training should be implemented as how-to-do-it or procedural learning for individuals with JNCL. The route should be considered as tasks within the activity it is attached to. For example, walking to and from the playground is the first and the last task in the activity «playing with friends at the playground». When individuals who are blind are navigating routes, the main features are landmarks, shorelines and laps.

Landmarks

A mobility route consists of two or more landmarks, identifying permanent topographical positions within routes, which may function as cues for travel and orientation. Landmarks can be objects such as a posts, curbs, fences or doors, or attributes of objects, for example the sound of a creek or the rough surface of a stone wall.

There are three basic criteria for selection of landmarks. The person must be able to detect and recognize the prominent physical features of the landmark, the landmark must be different from other objects in the area (e.g., a tree in the forest is not a useful landmark), and the landmark must not be moveable but have a permanent location.

Landmarks should have distinct characteristics and be easy to locate, identify, and distinguish from other objects in the environment. Good landmarks are prominent and appear as a change in the surroundings along the route. They must always be available when there is a need to make changes in the direction

of travel or indication of a hazard on the way, like traffic or steps. The distance between landmarks needs to be adapted to the attention span of the individual. For instance, the movements between two landmarks should not require more than ten seconds if the attention span – the time the person manages to keep the landmark in mind – is ten seconds. If there are not enough natural landmarks along the route, it may be necessary to create artificial ones, for instance, place a pole where the person is supposed to make a turn, place a carpet in front of the door to identify the classroom, or make a road bump on the sidewalk to indicate a danger of crossing traffic.

Tactile or haptic landmarks may be identified through touch and bodily movement. They may include posts, building corners, the end of a fence, and carpets on the floor, and should be identified and located without too much explorative efforts. These landmarks can be associated with almost any object or feature but cannot be detected from a distance. Objects that are too large are not suitable as tactile or haptic landmarks, because they usually do not function as distinctive points of references for effective orientation. It is important to avoid tactile or haptic landmarks that require too much cognitive effort and searching behavior, such as stretching or bending (Tellevik et al., 2007). Landmarks should be located in a position where they will be detected without unnecessary exploration, that is, on the shoreline that is used in the orientation.

One should try to select tactile/haptic landmarks that can be detected with the foot, hand or white cane, although taking into consideration that landmarks on the ground may disappear under snowy or rainy conditions. Tactile and haptic landmarks are in many cases perceived to be more stable and distinct than auditory landmarks. It is therefore recommended to use tactile/haptic landmarks when planning a new route for children and young people with JNCL who are blind.

Auditory landmarks may be sounds from a fan, a busy road, or a crossing signal on the traffic light. They may also be perceived as an echo from a wall or be related to the acoustics in a room. Objects that produce distinctive sounds when they are touched or hit can sometimes function as landmarks. Most auditory landmarks can be detected from a distance. However, a single sound from a car cannot be re-checked by the listener, and many auditory landmarks are not as permanent as tactile/haptic or visual landmarks.

A child who is blind may gradually change from tactile or haptic to auditory cues of the same landmark (or shoreline). Such a development should be considered a positive development, because it implies that the child has learned a more effective way of orientation, for example to use echo-localization.

Visual landmarks are objects or visual attributes of objects, such as size, shape, or color. Children with JNCL use visual landmarks in early childhood, but visual landmarks must be replaced with auditory or tactile/haptic landmarks after the

onset of blindness. From the start, one should try to select landmarks that can be used visually as well as tactile or haptic when planning routes for children with JNCL who have low vision. This may ease the transitions from visual to tactile/haptic cues.

Shorelines

Shorelines give the person continuous information along the way between two landmarks. Whenever possible, landmarks should be connected in pairs by a good shoreline, such as a wall, the edge between asphalt and gravel. There are tactile/haptic, auditory and visual shorelines. Tactile/haptic shorelines are preferred when planning routes for children with JNCL who are blind, because they are perceived as more stable and permanent than auditory shorelines.

It may be necessary to create artificial shorelines when there are no appropriate patterns along the route. For instance, one may build a rail between two landmarks or set up a sound beacon above a door. Further, like landmarks, an outdoor shoreline on the ground may disappear with snow or rain and might be replaced with a hand rail. Figure 16.5 illustrates the design of an artificial shoreline. The crosswalk has been raised, the person with JNCL can follow the raised edge of the crosswalk as a shoreline to allow for safe crossing.

Artificial shorelines do not have to be costly and providing them is rather a matter of finding functional and practical solutions. For instance, connecting a sidewalk and a playground located in an open field with a rope may enable a child with JNCL to play with other children without adult help.

Shorelines can be upright, such as handrails, or incorporated into the path, such as a pattern or surface texture. It is practical to select upright shorelines when possible. They are easy to follow and can be perceived also when there is snow or rain. Moreover, it is easier to follow upright shorelines like handrails with the hands than a pattern on the ground with the feet or white cane (Tellevik et al., 2007).

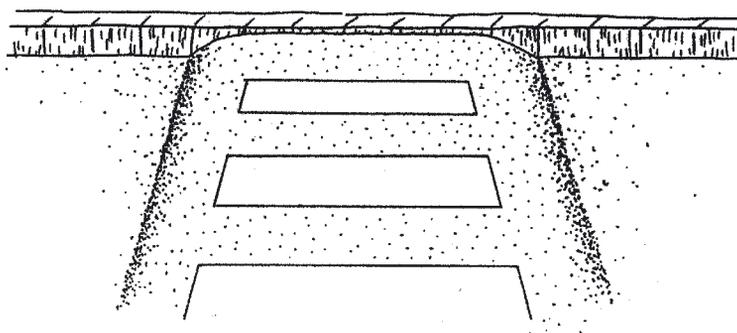


Figure 16.5
Example of physical
adaptation of a
shore line.

Laps

A mobility route can be divided into laps. A lap is the path between two consecutive landmarks in a route. To achieve independence, the person needs to learn all laps in a route. Learning one or several laps may be sub-goals towards achieving independence or interdependence in the route. The child may move according to interdependence between two landmarks by using free guiding.

There are two types of laps in a route. A *shoreline lap* is a path connected by two consecutive landmarks with a shoreline like a handrail, wall or curb. A *crossing lap* is the path between two consecutive landmarks that are not connected with a shoreline. Crossing laps are generally more difficult to learn than shoreline laps and should be avoided when possible. The first landmark in a crossing lap is often used for taking the right position and indicate the direction of the crossing. The second or last landmark in a crossing lap should be a landmark that intercepts the individual; the landmark should allow a margin of error of direction when crossing.

Learning and teaching laps and routes

Figure 16.6 shows an example of a route between home and school for a child with JNCL. The dotted line is the pathway and X1–X9 are landmarks. The landmarks are features such as beginning and end of a handrail, pillars and doors.

In Figure 16.6, "E" represents a full shoreline-lap with two subsequent landmarks (X3 and X4) connected with a shoreline. The child who is standing at landmark X3 (beginning of the handrail) can locate landmark X4 (end of the handrail) by following the shoreline (the handrail).

The lap from X4 (end of the handrail) to X5 (the edge between tarmac and grass) is a crossing-lap because it does not have a shoreline. The child with JNCL can use the handrail to find the right position for crossing the road. The child must uphold the direction given by the handrail while crossing the road to locate landmark X5. Maintaining correct direction while walking without a shoreline can be demanding for children with JNCL and should be avoided when possible.

Achieving independence in shoreline laps requires that the child or young person learns to locate the next shoreline (which is often attached to the previous landmark), the body's position in relation to the shoreline (left- or right-hand side), and the next landmark (attached to the end of the shoreline). The teacher's task is to direct the child's attention to these three tasks. The child can move independently in the lap when he masters these three tasks.

Achieving independence in crossing laps requires that the child takes the right position at the first landmark of the lap, for example by placing the back toward a wall or a handrail. The next step is to maintain the direction when crossing the open area. The last step is to locate the landmark that ends the crossing lap, for

instance, a wall or edge between tarmac and grass. The role of the teacher is to direct the child's attention to these three tasks, and the child will be independent when these tasks are mastered.

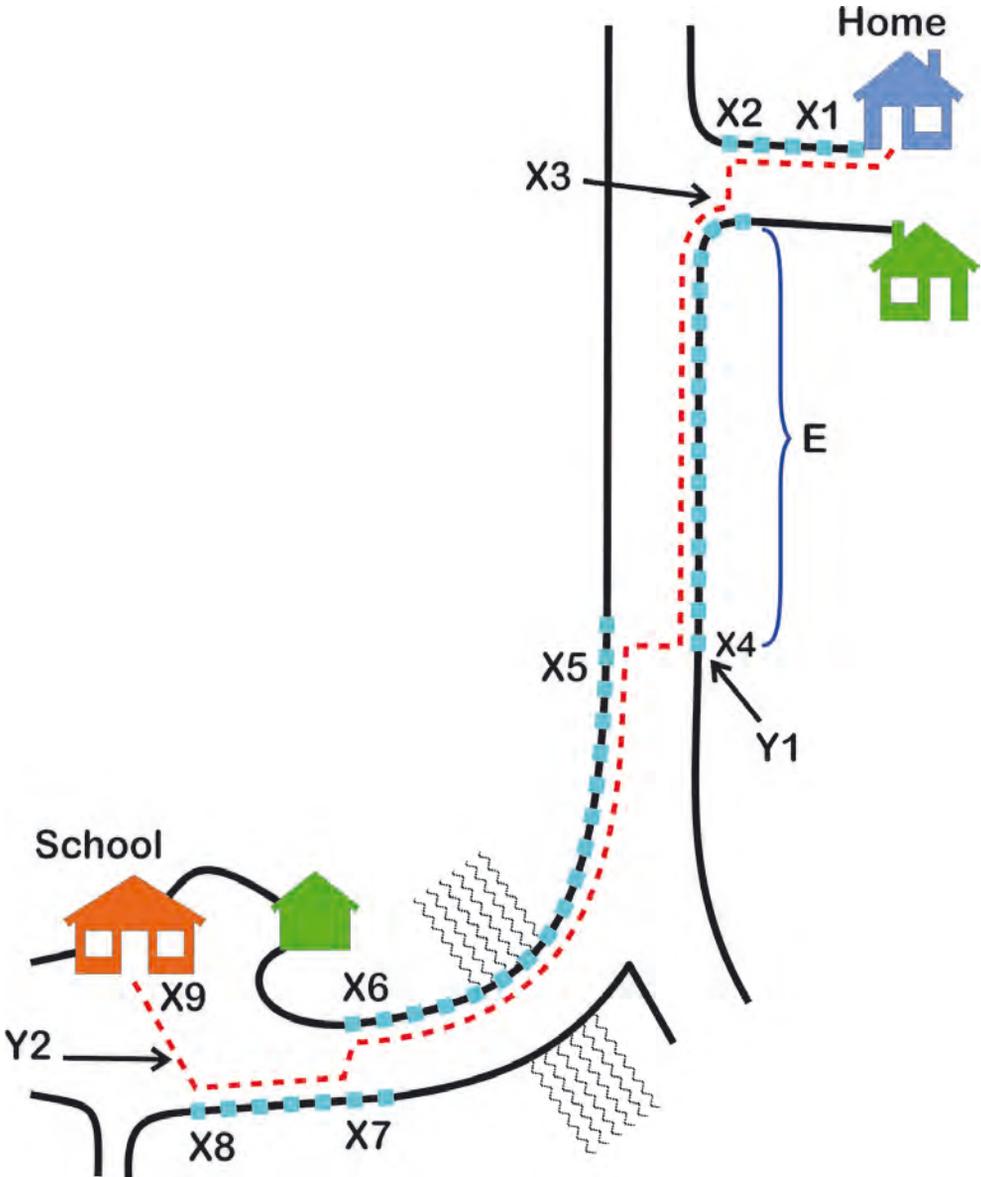


Figure 16.6 A mobility route between home and school for a child with JNCL. The dotted line is the pathway and X1-X9 are landmarks. Y1 and Y2 mark crossing laps and E shows a full shoreline lap (2 landmarks and a shoreline).

Planning routes

It is important to note that a route is a one-way pathway. In most cases it is not possible to use exactly the same landmarks and shorelines when returning to the starting point. The body position in relation to shorelines will always be on the opposite side when going back.

The teacher must always prepare the route prior to the training. This consists of selecting appropriate landmarks and shorelines and making physical adaptations in the environment where needed. An example of a prepared route from the classroom to the locker room is shown in Table 16.5 and the corresponding map in Figure 16.7. The dotted line in the map indicates the pathway. The route has 7 landmarks and 6 laps. Laps number 1, 3, 4 and 6 are shoreline laps while laps number 2 and 5 are crossing laps. The analysis disclosed a need for two physical adaptations, artificial landmarks.

The starting point of the route is the classroom door (landmark 1). By following the wall on the left hand side, the student locates the wall carpet (landmark 2). The wall carpet is a physical adaptation, this location had no features that could be used as a landmark or signal for the next crossing lap. The student takes position at the wall carpet (landmark 2), crosses the corridor and locates the wall on the opposite side (landmark 3). By following the wall on the right side, the student locates the corner (landmark 4), then turns right and follows the wall on the right side to the glass door (landmark 5). The student takes position at the glass door, crosses the corridor and locates the wall on the opposite side (landmark 6), and then follows the wall on the left hand side to locate the hanging rope (landmark 7). Landmark 7 is another physical adaptation, which enables the student to identify his own seat.

Table 16.5 The analysis of a route from the classroom to the locker room

ROUTE FROM CLASSROOM TO LOCKER ROOM			
	Landmark	Shoreline	Physical adaptation
1	Classroom door	Wall left hand side	
2	Wall carpet	No shoreline (crossing lap)	Wall carpet
3	Wall on opposite side	Wall right hand side	
4	Corridor corner	Wall right hand side	
5	Glass door	No shoreline (crossing lap)	
6	Wall on opposite side	Wall left hand side	
7	Shelf marked with rope		Hanging rope

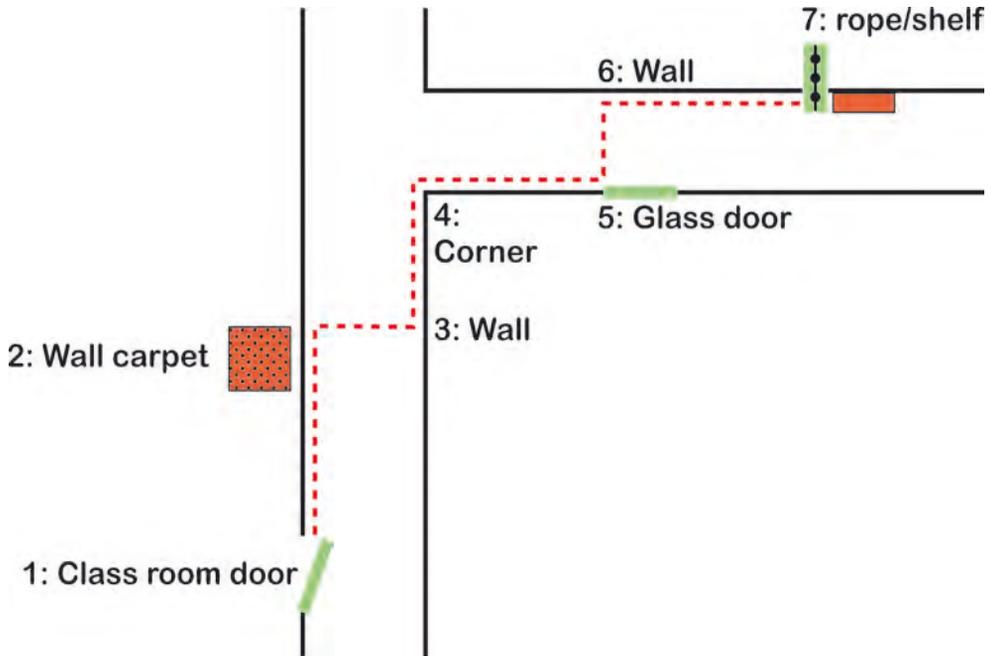


Figure 16.7 Map corresponding to the route in Table 16.5

Route training

Learning a route requires many repetitions of the predefined laps, landmarks and shorelines. The teacher's role is to guide the student's attention towards the pre-defined landmarks and shorelines on the route. The student must learn if the shorelines and landmarks are on the left or right side while moving, according to egocentric orientation. The teacher should not walk in front of the student when training independence, because the student may, without being aware of it, use sounds from the teacher for orientation purposes instead of using the predefined landmarks and shorelines. Unnecessary talking and information about things that are of minor importance for the orientation during the training may interfere with the student's attention towards the landmarks and shorelines. Every lap – from landmark to landmark – should be regarded as a sub-goal in the route training. One can strengthen the student's appreciation of laps or sub-goals by asking the student to stop at each landmark in the beginning of a training period.

Figure 16.8 shows an electronic form that can be used for training and evaluation of route training. The route describes the path from a classroom to the locker room, a distance of 190 meters. All the predefined landmarks are supposed to be signals for actions like «turn right», «cross over,» or «find next shoreline» based on egocentric orientation. The physical adaptations include artificial adaptations for making the orientation easier.

The JNCL and Education Project		Mobility route training			Erasmus+				
Name of student: Peter Jones		Name of teacher: Christine Smith		Date: 11/10/17					
From: Class room		To: Locker room		Route length: 190 meter		Trial no: 5		Time used: 2:25	
No.	Landmark	Shoreline - self to shoreline position and next landmark	Physical adaptation	Level of dependence					
				1	2	3			
1	Class room door	Follow wall on left hand to wall carpet		<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>			
2	Wall carpet	Cross corridor to wall opposite side	Wall carpet	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>			
3	Wall	Follow wall on right hand to corner		<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>			
4	Corridor corner	Follow wall on right hand to glass door		<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>			
5	Glass door	Cross corridor to wall opposite side		<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>			
6	Wall	Follow wall on left hand to rope		<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>			
7	Shelf marked with rope		Rope	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>			
8				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
9				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
10				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
11				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
12				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
13				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
14				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
15				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			
16				<input type="radio"/>	<input type="radio"/>	<input type="radio"/>			

Level of dependence to achieve the goal: 1 = no help from others, 2 = requires partial involvements from others, 3 = others have 100 % control

Figure 16.8 Form used for route training

The columns *Level of dependence* in Figure 16.8 is an evaluation of the student’s current orientation level on each lap. Level 1 indicates that the student can find the next landmark without help from the teacher. Level 2 indicates that the teacher must intervene in this lap, for instance by guiding the student to follow the shoreline or to locate the next landmark. Level 3 means that the teacher must take full control of the student’s orientation. The figure shows that the student with JNCL has achieved full independence in lap 1, 3 and 4, partial independence in lap 2 and 6, and needs full support in lap 5. The evaluation of lap 5 may indicate a need for further adaptation, for instance putting a carpet across the corridor which will change the present crossing lap into a shoreline lap.

A well-described route form is important when two or more teachers are training the same student. Route forms make it easier for staff to have the same expectations to the student and contribute to ensuring that they use the same laps, landmarks and shorelines.

Level of independence will over time be reduced for young people with JNCL. They will need more help when moving to desired activities. The earlier learned mobility routes may still be of importance despite less independence and more interdependence and dependence. Established mobility routes can be used for creating anticipation and maintaining personal overview and control over what is happening. For instance, landmarks like a door with a bell or a floor with

a thick carpet can make the student aware: *I am now on my way to the kitchen and lunch*. In later phases of the disease, use of fixed routes throughout the day is one method of supporting and maintaining the person's ability to understand what is going to happen next.

Guiding persons with JNCL

Sighted guides will become a necessity for persons with JNCL when the visual, motor and cognitive problems become more severe. Guiding is typically needed in unfamiliar environments, crowded places and places with certain hazards, or when there is a need to move fast. However, guiding should be avoided when not needed and it is an overall aim that students with JNCL should be as active as possible when they are guided. Active guiding means to use free guiding (see Figure 16.10) or verbal guiding, instructing students what to do rather than leading them with physical contact. Active guiding also implies letting the students open and close doors or pulling the chair out when they are sitting down.

The guiding grip

The person should hold the sighted guide as shown in Figure 16.9 when guiding by holding is needed. Children can hold the guide's wrist or fingers. The sighted guide should always walk a step ahead of the person with JNCL. There are several techniques that can be used when guiding by holding is needed (see Elmerskog et al., 1993; Jacobson, 2013; Wiener et al., 2010).



Figure 16.9 The guiding grip

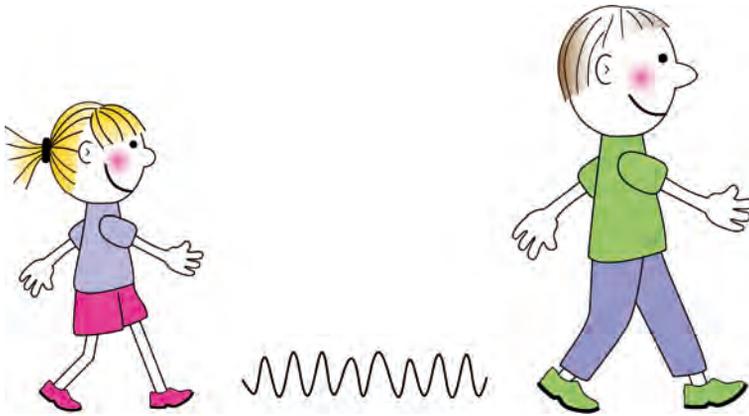


Figure 16.10
Free guiding

It is a risk that children with JNCL become overwhelmed by offers to be guided by holding just after the onset of the severe visual impairment. Adults and peers in the surroundings may think that they are supporting the child in best possible way when offering guiding. However, too much guiding may rather be a disservice, because the child may become accustomed to being transported or being a «passenger» when moving from place to place. The provision of guiding must be managed in a balanced way.

Free guiding

Free guiding should be given priority when guiding is needed for children with JNCL. Figure 16.10 shows an adult guiding a child with JNCL. There is no physical contact between them, the child uses sounds from the guide as cues for her orientation. The guide may make sounds with the feet, by talking to the child or by holding a device that makes sounds. Guidance from a distance requires more attentional resources from the student, compared to guiding by holding.

Free guiding may be a step towards later independence. It is therefore recommended to use designed routes when free guiding is used, because the child may memorize laps, landmarks, shorelines and turns on the way to the destination.

Guiding wheelchair users

Using designed routes may also be important for a person with JNCL using a wheelchair when moving from place to place. Perceiving 90 degrees swings at the same location every day and passing the same uneven and rough surfaces, carpets or doorsteps may establish a foundation for later orientation. Also changes in speed can be used for orientation. For instance, a pronounced 90 degrees turn combined with a door still may be recognized as the entrance to the school

canteen. Guiding a person with JNCL in a wheelchair should always be combined with verbal information about where to go, features in the surroundings, and that the destination is reached. A person with JNCL in a wheelchair should always be placed close to known and recognizable physical features and objects, such as behind a known desk or beside a familiar wall while sitting still.

Suggestions from parents and staff

Comments from parents and staff in the present JNCL and education study (Appendix A) included several suggestions that may be useful for enabling children and adults with JNCL to cope in daily life. Some of these were related to self-care and grooming. Containers with soap or shampoo can be marked with braille, Moon, a rubber band or small object taped to the container. Identification will be easier with different sizes and shapes of bottles and tubes. It is easier to

get a suitable amount of soap with a pump bottle, or to press out the right amount of toothpaste by squeezing it directly into the mouth from personal tubes.

Other suggestions were related to dressing and undressing. Clothes should be easy to handle for the young person when the disease progresses. For instance, shoes with Velcro fastening may maintain independence when shoes with laces become difficult to handle. Short moments of training in natural situations may provide a basis for automatizing skills and developing fine motor skills such as using a zip, buttons, or laces. Putting the clothes in a certain order when undressing makes dressing easier (see Figure 16.11). Parents should take the prevailing youth styles, dress codes and the child or young person's own voice into account



Figure 16.11 Systematic organization of clothes

when choosing or purchasing clothing. A mother told that her daughter was obsessed with having a hair braid, a style that was very popular prior to the daughter's visual impairment. The fact that this hair fashion had faded was never communicated to the daughter, and her obsession with hair braids disappeared when she was told about the new hair trends.

It is advantageous to practice skills related to eating and drinking from early childhood. The child can take part in preparing food and learn hand-over-hand to chop vegetables or butter a slice of bread. Pouring drinks can be learned with practice, as well as laying the table, taking food from the kitchen and carrying it to the table, or taking the dishes to the dish washer after dinner. Such skills can be accomplished independently or through interdependence. As skills decline, eating becomes more difficult. Physical adaptation may include putting a supplementary edge on the plate, using thicker cutlery, or putting an anti-slip pad under the plate.

The disease may make swallowing more difficult, and with age feeding the person with JNCL may be needed. In such cases the person should be told with speech or touch when the food is just about to enter the mouth. Severe problems with swallowing may require chopped or liquidized food. Drinking may become easier if the liquid is thickened.

Many parents mention that the use of money is an inspiring activity for many children with JNCL. A wallet with money or a card and paying for purchased items in the shop can be used for building up an understanding of money values and mathematics in general. A coin holder can be used for storing money, which also makes the differentiation of coins easier.

Parents in the present study reported sleeping problems for two thirds of the participants with JNCL, at a mean age of onset of 11 years. The need for rest and sleep will increase but it is important to plan for activity and participation, and to a lesser extent for rest and sleep, although considering the need when it becomes comprehensive. Sleeping problems can be met by medication, but the establishment of best possible daily rhythm is as important as for most people. The daily rhythm should be regular and contain relatively few changes. Quiet routine activities before bedtime, like listening to an audio book or soft music, may make it easier for the person to fall asleep. The best possible daily rhythm is a matter of being energized and engaged in appropriate activities through personal involvement.

Parents and staff reported that nightmares, confusion in connection with sleeping and waking up were increasing with age. Some emphasized the importance of immediate contact with helpers or other adults after waking for stopping further escalation of confusion and desperation. However, falling asleep during daytime is much more common when not being engaged or involved in activities.

Some persons with JNCL may take instant naps while being involved in activities. According to parents and staff in the present study, long-lasting activities should be avoided when possible for persons with short attention span; variation and short-lasting activities are of help to maintain spirit and engagement.

References

- Agich, G. (2003). *Dependence and autonomy in old age: An ethical framework for long-term care*. Cambridge, UK: Cambridge University Press.
- Aro, T., Siiskonen, T., Niemelä, R., Peltonen, M., Stenroos, H., & Kulmala, E. (2007). Opettaminen ja oppiminen (Teaching and learning). In T. Aro, T. Ahonen & T. Siiskonen (Eds.), *Ymmärsinkö oikein? Kielelliset vaikeudet nuoruusiässä* (Did I understand correctly? Linguistic difficulties in adolescence) (pp. 113–118). Jyväskylä: PS-Kustannus.
- Baltes, P. B. (1997). On the incomplete architecture of human ontogeny: Selection, optimization, and compensation as foundation of developmental theory. *American Psychologist*, *52*, 366–380.
- Baltes, P. B., & Baltes, M. M. (1990). Psychological perspectives on successful aging: The model of selective optimization with compensation. In P. B. Baltes & M. M. Baltes (Eds.), *Successful aging: Perspectives from the behavioral sciences* (pp. 1–34). Cambridge: Cambridge University Press.
- Csikszentmihalyi, M. (1997). *Finding flow: The psychology of engagement with everyday life*. New York, NY: Basic Books.
- Elmerskog, B., Martinsen, H., Storliløkken, M., & Tellevik, J. M. (1993). *Førhøretsoplæring – Mobilitet i en funksjonell sammenheng* (Mobility training in a functional context). Trondheim: Tapir.
- Emerson, R. W., & McCarthy, T. (2014). Orientation and mobility for students with visual impairments: Priorities for research. *International Review of Research in Developmental Disabilities*, *46*, 253–280.
- Entwistle, V. A., Carter, S. M., Cribb, A., & McCaffery, K. (2010). Supporting patient autonomy: The importance of clinician-patient relationships. *Journal of General Internal Medicine*, *25*, 741–745.
- Ferrara, K., & Landau, B. (2015). Geometric and featural systems, separable and combined: Evidence from reorientation in people with Williams syndrome. *Cognition*, *144*, 123–133.
- Fraiberg, S. (1977). *Insights from the blind*. New York: Basic Books.
- Gibson, J. J. (1966). *The senses considered as perceptual systems*. Boston, MA: Houghton Mifflin.
- Giebel, C. M., Sutcliffe, C., & Challis, D. (2015). Activities of daily living and quality of life across different stages of dementia: A UK study. *Aging and Mental Health*, *19*, 63–71.
- Greenfield, P. M. (1994). Independence and interdependence as developmental scripts. In P. M. Greenfield & R. R. Cocking (Eds.), *Cross-cultural roots of minority child development* (pp. 1–40). Hillsdale, NJ: Lawrence Erlbaum.
- Jacobson W. J. (2013). *The art and science of teaching orientation and mobility to persons with visual impairments, Second edition*. New York: AFB Press.
- Lancioni, G. E., Sigafoos, J., O'Reilly, M. E., & Singh, N. N. (2013). Orientation systems for promoting indoor travel. Chapter 5 in *Assistive technology* (pp. 107–127). New York: Springer.
- Lowenfeld, B. (1954). Effects of blindness on the cognitive functions of children. *The Nervous Child*, *7*, 45–54.

- Martinsen, H., Tellevik, J. M., Elmerskog, B., & Storliløkken, M. (2007). Mental effort in mobility route learning. *Journal of Visual Impairment and Blindness*, 101, 327–338.
- McConachie, L. (2018). School readiness and kindergarten transitions: Children with vision impairment and blindness. In A. J. Mashburn, J. LoCasale-Crouch & K. C. Pears (Eds.), *Kindergarten transition and readiness* (pp. 205–223). Cham, Switzerland: Springer.
- Mulloy, A. M., Gevarter, C., Hopkins, M., Sutherland, K. S., & Ramdoss, S. T. (2014). Assistive technology for students with visual impairments and blindness. In G. E. Lancioni & N. N. Singh (Eds.), *Assistive technologies for people with diverse abilities* (pp. 113–156). New York, NY: Springer.
- Newcombe, N. S., Uttal, D. H., & Sauter, M. (2013). Spatial development. In P. Zelazo (Ed.), *Oxford handbook of developmental psychology, Volume 1* (pp. 564–590). New York, NY: Oxford University Press.
- Prizer, L. P., & Zimmerman, S. (2018). Progressive support for activities of daily living for persons living with dementia. *The Gerontologist*, 58, S74–S87.
- Rogoff, B. (1990). *Apprenticeship in thinking – Cognitive development in social context*. Oxford, UK: Oxford University Press.
- Taylor, J. S. (2005). *Personal autonomy*. Cambridge, UK: Cambridge University Press
- Tellevik J. M., & Elmerskog, B. (2001). *The mobility and rehabilitation programme in Uganda. A sociocultural approach to working with visually impaired persons*. Oslo: Unipub.
- Tellevik, J. M., Storliløkken, M., Martinsen, H., & Elmerskog, B. (2007). *Spesialisten inn i nærmiljøet* (The expert in the proximal environment). Oslo: Unipub.
- Törrönen, S., & Onnela, J. (1999). Vapaus tulla, vapaus mennä, Sokean ja heikkonäköisen liikkumistaito ja sen kehittäminen (Freedom to come, freedom to go! Blind and partially-sighted mobility and development). Arlainsituutin julkaisuja 2/99. Saarijärvi: Gummerus Kirjapaino Oy.
- Vasilyeva, M., & Lourenco, S. F. (2012). Development of spatial cognition. *Wiley Interdisciplinary Reviews: Cognitive Science*, 3, 349–362.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- von Tetzchner, S., Fosse, P., & Elmerskog, B. (2013). *Juvenile neuronal ceroid lipofuscinosis and education Biochimica et Biophysica Acta (BBA)-Molecular Basis of Disease*, 1832, 1894–1905.
- von Tetzchner, S., & Martinsen, H. (2000). *Augmentative and alternative communication*. London, UK: Whurr/Wiley.
- Vygotsky, L. S. (1962). *Language and thought*. Cambridge, MA: MIT Press
- Wiener R., W., Welsh R. L., & Blasch, B. B. (2010). *Foundations of orientation and mobility, Third edition: Volume 2 Instructional strategies and practical applications*. New York: AFB Press.
- Zhou, L., Parker, A. T., Smith, D. W., & Griffin-Shirley, N. (2011). Assistive technology for students with visual impairments: Challenges and needs in teachers' preparation programs and practice. *Journal of Visual Impairment and Blindness*, 105, 197–210.

Music Education and Music in Education

Adam Ockelford and Rebecca Atkinson

This chapter is about the role of music in the education of children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL). The survey presented in this book (Appendix A) is the first systematic study ever to be undertaken in this area, although anecdotal accounts of parents, and reports of teachers and music therapists, have in the past suggested that music may potentially have an under-utilized role in the lives of individuals with juvenile neural ceroid lipofuscinosis (JNCL) (Bills, Johnston, Wilhelm, & Leslie, 1998; Gylfason, Jóhannsdóttir, & Einarsdóttir, 2006; Heikkinen & Railosvuo, 2006; Olsén & Laine, 2006).

Music, whether heard or performed, is something that is experienced through the body and in the mind, and research has shown that the brain can continue to respond to music even when there is damage or degeneration (Baird & Samson, 2015). While music uses certain specialized areas in the brain, there are close connections too with the neurological resources devoted to language, movement, memory and emotion, and it has been suggested that the existence of these connections may be central to the efficacy of music interventions (Woods et al., 2005).

Main results from the survey

Parents were asked about their child's use of music and what impact the music had had on the life of the child. Similar questions were asked to professionals working with children and young people with JNCL. The informants were asked at what age the children began to engage with music, and for how long this engagement continued. There were questions too concerning music lessons and music therapy sessions, and about the potential power of music to facilitate social contact, to support communication, to provide comfort, to aid learning and understanding, and to offer stimulation and relaxation.

Most of the children and young people in the present study were said to be interested in music, whether as listeners or performers, or as an accompaniment to dance or a topic of conversation. In the survey, 87.4 percent of the parents said that music had or once had a high or very high impact on the life of their child. While the importance of many activities was declining with age, the percentage who found music important increased from around 36 percent in the age bracket 7–9, to around 50 percent at ages 13–15 and 16–21, and nearly 80 percent among those above the age of 22 (Figure 17.1). These substantial changes from childhood to adolescence and then young adulthood are probably a reflection of the fact that the enjoyment of music does not rely on intellectual capacity or motor abilities. Similarly, the impact of audio books, another receptive activity, was said to increase gradually from less than ten percent at 7–9 years to over 50 percent above the age of 22 (see Chapter 14).

Susan always had her music with her after she started to go blind. Up to the end music was a big part of her life.

The active use of music, such as performing and dancing, was most prominent in childhood and early adolescence. Singing was said to be at its best (and on a par with sighted peers) around the age of 12.

I think that song and use of the voice is an instrument. He sang a lot up to the time when his voice began to break. The decline of skills started to accelerate during puberty.

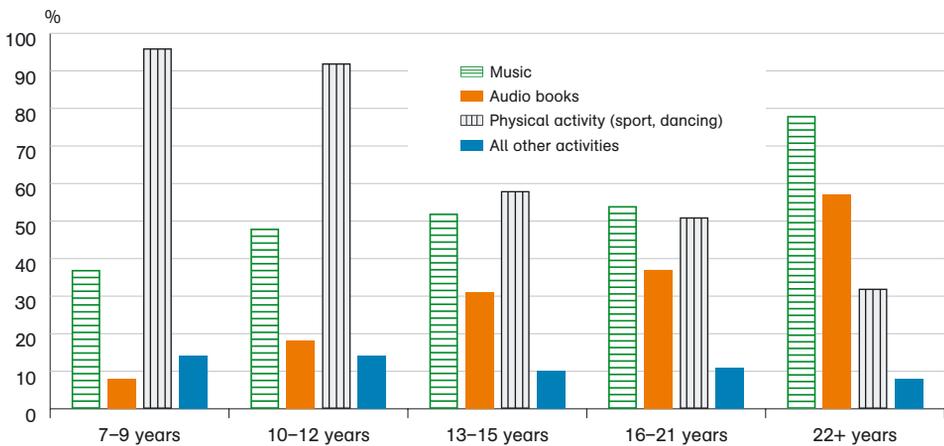


Figure 17.1 Percent of participants for whom music, audio books, physical activities and all other activities were important at different age levels

She used to have a very nice singing voice, but that has disappeared as the disease has progressed. She still enjoys singing, although it is usually just the last word of each line.

Around half of the children and young people with JNCL played, or had once played, an instrument, and some had tried two or more instruments. The most popular instruments were the keyboard (including the piano), drums (or other percussion) and the guitar.

Drums and drum kit. Started to play around 6–7 years of age. Achieved best skills (the peak) around 12–13 years of age. Played in a band organized because of him, he continued doing this up to 20 years of age. He was also offered special training (one-to-one).

Most children started playing instruments around the age of eight (some earlier), which continued for around five years. Around two thirds (65.5%) did not get beyond the level of beginners, while the rest were described as performing at a similar level to or better than their peers.

Since the age of 4–5 years (7 now), he wanted to play and own every instrument. He took drum lessons for eight months and guitar for two months when he was six and seven. He is neither technically trained nor proficient, but he has great rhythm and musicality. He has played or plays the following: drums, cowbell, cymbals, triangle, washboard, harmonica, recorder, spoons, guitar (acoustic and electric), clarinet, trumpet, violin and keyboard.

It seems that the children started to engage more with music as their eyesight deteriorated, but generally stopped when they could no longer read print music notation. Only one adolescent was reported to have attempted to learn to read music in braille. Around 20 percent were reported to make up (or to have made up) their own music. This occurred largely between the ages of 11 and 15.

Around one third of the parents (32.1%) said that their child had received music therapy. The median age for starting was 14, and the mean duration of the music therapy was eight years. Within this group, around 90 percent of parents and 80 percent of professionals reported that music therapy had (or once had) a high or very high impact – primarily as a source of stimulation and comfort.

The potential function of music for children and young people with JNCL

Sixty-nine parents commented on the potential role of music in support of wider functioning and wellbeing. Many said music helped their child to sustain communication when words alone no longer functioned, that music was a medium through which the child could convey feelings when other channels of emotional expression were occluded. Parents also reported that music reawakened memories that otherwise appeared to be lost, and how music helped their child to maintain a sense of wellbeing in an increasingly confusing and frightening inner world.

This section discusses the findings from the survey in relation to these issues, compares them with those from other research, and makes suggestions for musical activities and strategies that may be useful for families and for professionals working with individuals who have JNCL or other comparable disabling conditions.

Feelings

Just under half of the respondents identified music as being important in promoting individuals' wellbeing, assisting with emotional regulation and expression, and relaxation. This is particularly important for individuals with JNCL since they experience multiple losses – in skills, friendship and the availability of activities in which they can participate – which can cause anxiety, uncertainty and anger.

Music was very important to her. She liked music for relaxation, as well as music for singing along (which was functioning for an astonishingly long time). Music also evoked strong emotions, grief as well as joyfulness. Music therapy at school was one of her favorite subjects. She always liked music while being together with others, at school, with the family or on organized vacations. Music brought about a high level of wellbeing.

She played her instruments, guitar and drums, and she enjoyed singing. Soft music was used to calm her down the last years she was alive (to prevent anxiety and restlessness).

To manage such feelings, one parent described how «music could change a child's mood and make it better», sometimes through bringing back good memories. Another parent reported how playing the drums helped her child maintain his self-confidence: *music has been, and still is, his greatest passion*. Similarly, a professional noted that music could both produce *a high level of excitement* as well as having *a calming effect during critical situations*.

Using music to create a feeling of calm can be particularly important at bedtime: one family used it when their daughter was going to sleep *to create a feeling of unity*, acting as Ockelford (1998) puts it, as an auditory "security blanket." A number of parents and staff commented on how music was used to distract or as a diversion during moments of personal care, to reduce anxiety in everyday activities like travelling in the car, and to help with pain management. Whatever the circumstances, *preferred* music was said to be the key: Bills and associates (1998, p. 14) reflect that children with JNCL often have «favorite music ... that they seem to hang onto and will listen to over and over.»

Olivia's story

Olivia is 15 years old and lives in the UK with her mother, father and two brothers at home. She attends a school for children with special educational needs, to which she moved just over a year ago from a mainstream secondary school. Olivia is chatty and sociable, and becomes animated when talking to others about her passions, preferences and experiences. She loves music, and among her favorite pieces are songs from Disney, Westlife, Britney Spears and Rihanna.

Olivia says that she enjoys being at school, and is «always happy». However, when she first moved to her new school, this was not the case, and staff reported that Olivia suffered a good deal of anxiety, clutching at the walls of the corridors, apparently terrified of being in unfamiliar surroundings. It was suggested that music could be used to help her relax as she got used to her new school environment, and weekly music therapy sessions were arranged with a clinician at school. Staff commented how these sessions lifted her mood.



Olivia.

Olivia has significant difficulties with speech. She talks very quickly, often mis-accentuating words or stammering, and repeating things in an effort to be understood. However, when Olivia is singing along to her favorite music – for example, in music therapy sessions – her capacity to use words is transformed. She sings in time, with linguistic clarity and precise articulation. She maintains

the contours of melodies, though not the precise pitches of each note. But the important thing is that she is able to produce language that others can understand.

Language and communication

As the disease progresses, children and young people with JNCL increasingly face challenges in the production of language: speech increases in speed, with rapid stuttering; articulation is impaired and words are mispronounced (see Chapter 6). Eventually, the speech may become incomprehensible. However, the capacity to sing appears to remain intact – sometimes for years – beyond the point at which speech is difficult or impossible to understand, and this was reflected in a number of the observations made by parents and professionals. For example, a child was reported to speak more clearly when singing what she wanted to say, and another recognized that the *singing of texts is better than speaking*.

Music was very important – she enjoyed listening and singing. Long after her speech went she was still able to sing or mouth the words to “Happy birthday to you” for example.

One young person was said to really enjoy listening to her favorite artists and songs throughout the day. She also went to concerts. Singing clearly brought her pleasure, and in her singing the *lyrics came out clearly, even though her speech was so little, stuttering and slow*.

Why should this be the case? Research extending back over several decades has suggested that in songs, music and language may be encoded together in the brain (e.g., Morrongiello & Rose, 1990; Serafine, Crowder, & Repp, 1984), findings that have found support in more recent neuroscientific work (e.g., Schön et al., 2010). According to Patel (2007) elements of linguistic and musical processing may share some resources in the brain, while nonetheless having areas that are discrete. Hence one may speculate that in conditions such as JNCL, when the "direct" route to speech production becomes damaged, an "indirect" route remains open, through singing. The "shape" or "contour" of a song's melody (its ups and downs) and its rhythm (pattern of accents over time) may open the door to song production. The effect is that the natural prosody of language – its "musical" qualities – are exaggerated as strings of words are produced, but the sense is clear.

Despite this, little evidence emerged from the present survey that music was being used systematically to support, enhance or maintain speech, although one child was reported to be taking singing lessons to stimulate speech production. However, work in the UK, developed in the 1980s at Linden Lodge School in London, with children with visual impairment and additional disabilities, including



Figure 17.2 Songs for social expressions

children with JNCL, pioneered the use of "micro-songs" – key words and phrases that were set to short melodies (Ockelford, 1994) (see Figure 17.2). These were taught to children in the course of music sessions as part of social games, and children were subsequently encouraged to transfer them to real life situations to be used functionally. In some cases the teachers supported the production of words by children who were in the early stages of speech, or had acquired articulatory difficulties through brain injury or disease. In other cases, the micro-songs served as a proxy for words – for children who were unable to make speech sounds but could nonetheless vocalize. Here, the rhythm and, in some cases, the contour of the melody, was all that was required for the meaning to be clear. Key factors in the success of this use of music were that all those involved in the education and care of the children concerned were aware of the songs and their potential meanings, and that teachers and others were prepared to employ them consistently themselves, to model and reinforce their use (Ockelford, 1996). Here, longer songs incorporated key words and phrases that consistently used the same melodic shape and rhythm – characteristic motifs that could be extracted for deployment in everyday situations (see Figure 17.2)

There are many anecdotal accounts that young people with JNCL – like other young people – enjoy singing and music, but there is hardly any research on the potential impact of music therapy and education in this group. Phase I of a new UK research project termed *Music in Neurodegenerative Disease* (MIND) is examining the potential efficacy of music to support communication and thereby enhance the life of young people with JNCL.¹ The project includes the creation of a library of 39 potential "micro-songs" and Figure 17.3 shows how these are organized conceptually. This scheme is currently being piloted with children and

1 Sponsored by the Baily Thomas Charitable Fund, and supported by The Amber Trust (www.ambertrust.org), and the subject of doctoral research by the second author.

young people with JNCL. It is anticipated that each person will use only a small selection of micro-songs to help with communication, and will start with the core. New songs will almost certainly need to be developed for individuals, based on preferences and needs.

The approach set out here is similar to techniques used in an intervention called Neurologic Music Therapy. These include Melodic Intonation Therapy (MIT) and Music Speech Stimulation (MUSTIM) (summarized in Thaut, 1999). MIT aims to facilitate spontaneous speech through sung and chanted melodies that resemble speech intonation patterns. The step-by-step process should be used over an extended period of time, and it may be helpful for family members as well



Figure 17.3 Map of "micro-songs" used in *All Join In 2*

as music therapists to adopt the approach to encourage expressive communication. MUSTIM uses musical materials such as songs, rhymes and chants to encourage language production through the completion of learned familiar song lyrics, association of words with familiar tunes, or musical phrases. For example, spontaneous completion of familiar sentences is stimulated through familiar tunes or obvious melodic phrases. It can be used as a follow-up to MIT, as an attempt to increase the number of functional spoken utterances that the individual is able to produce. These techniques are widely used by speech and language therapists and neurologic music therapists when working with individuals who have various forms of neurological impairments (see for example www.chilternmusictherapy.co.uk).

Music and movement

Music is related to movement in two important ways, both of which pose challenges and afford opportunities for children and young people with JNCL. First, hearing music naturally stimulates movement; indeed, some cultures do not separate the notion of "music" from the concept of "dance" (Lewis, 2013). Second, movement (of some kind) is invariably required to make music, whether through singing, playing an instrument or using specialist technology.

The close connection between music and movement can be used to assist those with JNCL, for whom physical activity of any kind becomes increasingly difficult as the disease progresses. For example, one parent noted that *walking ... succeeds when she is listening to her favorite music and singing along* and another parent commented that music helped her son to be more coordinated and aware of his body. Although formal research into the capacity of music to function in this way for young people with JNCL has yet to be undertaken, it seems likely that some of the specialist techniques of Neurologic Music Therapy can be of benefit. For example, rhythmic auditory stimulation (RAS) draws on the brain's ability to entrain to the underlying rhythmic regularities that most music exhibits – enabling people to move the body in time with the beat of the music.

A number of songs from *All Join In!* (Ockelford, 1996) were designed to encourage and offer a framework for movement in blind children with other disabilities, and these have been extended in the MIND project. They include music to walk, rock, stretch and relax to. The titles of these songs appear in the upper left quadrant of Figure 17.3.

Producing music and sound is a sensorimotor experience that can be both felt and heard, and one that requires co-ordination and control when using musical instruments (Ockelford, 1998). For example strumming a guitar or pressing the keys of a piano demands finger dexterity and strength, while beating drums

and cymbals requires large upper or lower body movements to create sounds. Vocalizing, singing or blowing instruments regulates breathing and encourages people to exercise breath control and strengthen their respiratory system. Whatever movement that is required, the driving force is intrinsic motivation: the will to move the hand derives from the desire to make a certain sound.

The challenge for professionals working with young people with JNCL is how to offer them access to music-making as physical coordination and strength diminish. One option that is being explored in the MIND project is to use MIDI-based technology through which switches and beams can be used to control sounds generated by means of a computer (see Chapter 19). This means that even the tiniest movement can be used to trigger a whole orchestra of instrumental possibilities. In terms of research, currently the most relevant in-depth study pertains to Abby, a young woman in the late stages of juvenile Tay-Sachs disease (Ockelford, 2012). In the nature of its progressive mental, physical and sensory decline, Tay-Sachs is very similar to JNCL – indeed, the two were often confounded in the past (Walkley, Siegel, & Dobrenis, 1995).

Abby's story

Abby's illness had started to take effect when she was two years old. By the age of seven, she had lost the ability to speak, although she was still able to sing the words in songs for some time afterwards. The challenge of Abby's music teacher in the last stages of her illness, when she was unable to vocalize or grasp a conventional musical instrument in order to manipulate it, was how to enable her to engage proactively in her class music sessions. He tried a technological approach, using the OptiMusic® system. This works by emitting beams of light, which, when broken at different points by a reflective surface, are detected by a sensor, and cause corresponding MIDI signals to be sent to a digital library of sounds. The reflective surface was provided by an ultra-lightweight plastic paddle, which, with some preliminary assistance, Abby could grasp with her right hand. It initially rested on her lap, and the beam was calibrated so that a movement of only two or three centimeters was sufficient to trigger a note. The system was set up so that Abby had a melodically pleasing series of pitches available to her (C, E and G). The music teacher led the supporting staff in improvising a song that used Abby's notes, and provided a keyboard accompaniment. The results were remarkable. Having been unable to produce music herself for a number of years, in only a short space of time, Abby showed that she was indeed able to participate in music-making, and produced sounds with increasing vigor as the song progressed over a couple of minutes. Most surprising of all, she could not only keep in time with the beat, but she was able to imitate motifs from the tune that the staff sang (her achievements can be seen at www.soundsofintent.org).

Music to mark time and provide information about activities, places and people

In the present survey, a number of parents and professionals mentioned that music was used throughout the day to mark out key events, and to provide emotional succor at potentially difficult times. For example, one young person with JNCL had *set songs for going to the toilet and brushing her teeth*. Another invariably listened to music in the morning before school, to ease the transition.

Music is used every day. He has special songs to fall asleep to; different songs have been used in different situations (pee song, wake up song, be together song).

Beyond this group, work with children who are blind and have learning difficulties over the last three decades has shown how music can be used systematically to structure the environment using auditory support, such as adding clearly defined and consistent sounds to rooms and other areas to compensate for the lack of visual information available to the individual (Ockelford, 1998, 2008). For example, brushing past a particular set of wind-chimes may indicate entry to a pupil's classroom; the same relaxing music may always be played on entry to the multisensory room; and a tuneful door-chime may be used to remind the individual that he or she has come home. At the same time, it is important to reduce "auditory clutter" that may function as distraction: classrooms can be very noisy places! Sometimes, moving to a smaller, quieter space may facilitate the pupils' concentration.

Key activities may be initiated using sounding objects: a small bell for music, for example, a larger one for gym, or the rattle of a chain for an outdoor play area. Such objects can be used to make "auditory timetables", so that students can be helped to anticipate a morning's activities, and can reflect on them once they are past – perhaps by placing the objects in a series of boxes whose lids are closed, in order to indicate completion. The identity of people too can be enhanced through the use of different sound-makers such as jangly bracelets, as well as micro-songs (see Figure 17.3).

Music to encourage socialization – structured and informal

Engaging with music, whether as a listener or active participant, offers an interest that all young people can share with their family and friends. Developing strong musical preferences in adolescence, and sharing these with peers, is a normal part

of growing up and developing an identity that is at once unique and indicates belonging to a group of like-minded peers. The comments in the survey suggest that this is just as true for young people with JNCL as it is for young people without disabilities. For example:

My daughter has loved music all her life and still does. Singing was very important to her and sharing music with friends; she used to want to record music on cassettes to give to her girlfriends ... it was a social interaction too.

From an educational perspective, one professional observed:

Through the morning gatherings with music every day at school, the child has shown development both socially with other pupils and has been able to communicate.

However, the parents cautioned about possible dangers. One parent emphasized how music could be misused, as a substitute for social contact, which had a detrimental effect on the care of her son with JNCL.

Music has been something he has been good at since he was a child, and it gives him a lot of pleasure. But it can be abused by the carers in the sense that they think they can just play him a CD instead of spending time and communicating with him. He likes being talked to by the carers, even if he doesn't verbally respond. It's a really big challenge with a lot of unskilled workers.

Some proprietary music technology programs also offer an effective way of engaging some individuals in music-based projects with their friends or peers, although using these independently in the absence of vision can be difficult. Nonetheless, when working as a group with sighted peers, young people with JNCL may be able to choose and build up loops, beats and sounds, and contribute to song-writing activities by simply indicating their choices through words, vocalizations or gestures. Other forms of song-writing are possible too, in which the song words are changed to relate to specific interests or topics that are relevant to the individual with JNCL.

Music can also be used in more formal educational and therapeutic contexts to facilitate social contact, scaffold interaction, promote turn-taking and listening to others, and to promote participation. Examples are outlined in Figure 17.4.

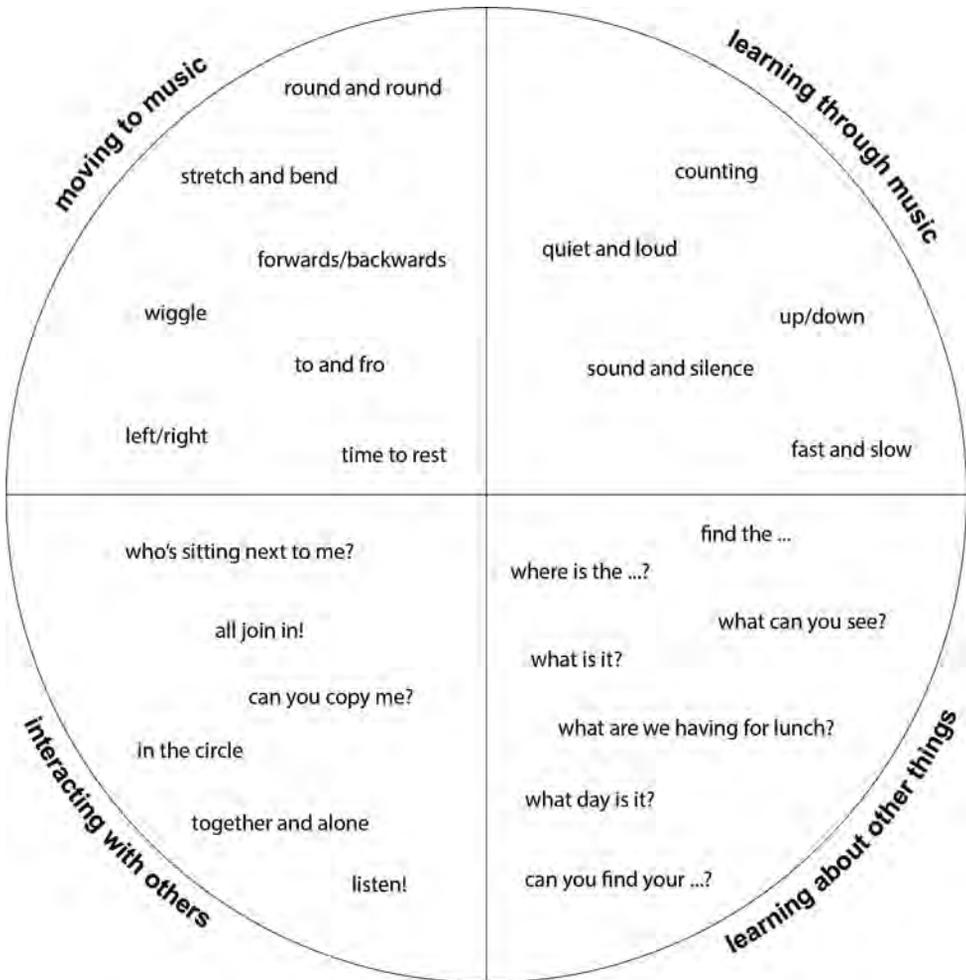


Figure 17.4 Map of songs to encourage wider development, understanding and wellbeing used in *All Join In 2*

Laura's story

Laura is 18 years old and lives on a supported campus in the UK, which is specifically set up to support young adults with JNCL. However, she regularly sees her family for trips, overnight stays and when attending church together.

Laura has a passion for music and enjoys listening to her vast music collection in her spare time. She particularly enjoys Disney songs, Avril Lavigne and Leona Lewis. She has music therapy sessions each week, and brings with her a large folder full of music for the music therapist to sing and play with her. As with many other young adults with JNCL, Laura's working memory is poor, and she often asks

the same questions repeatedly in order to confirm events that have happened recently or may happen in the near future. Her long-term memory appears to be more intact, and in therapy sessions Laura has an almost errorless memory for singing familiar songs, and for melodies that she used to play on the piano as a child. She will even correct the music therapist regarding the exact number of choruses in each song.

In order to sustain and nourish these memories, the music sessions focus on using both familiar and improvised music. During one session, Laura was

taking great delight in beating on a large drum to the words "a crash of drums" and softly sweeping the wind chimes to represent "a flash of light" during a rendition of the song *Any dream will do* from *Joseph and his amazing technicolor dreamcoat*. Through this interactive activity she was creative, playfully painting the song with musical sounds. This led Laura to think about how music and instruments can portray other stories, discussing what sound and music could be used to depict the characters and actions within them.

The music therapy sessions provided Laura with a motivating context in which to rehearse skills that might help in her everyday life. By engaging in activities that involved choosing instruments, sounds and songs, Laura was encouraged to maintain her ability to choose and to communicate these choices – allowing her voice to be heard. Through song-writing activities, Laura was given the opportunity to create new materials, involving verbal comprehension, planning, organizing and problem-solving. Put simply, music was the mainstay of Laura's life.



Laura.

Music and memory

Music can fulfil a special function in helping to revive memories. For example, one professional reported that *familiar pieces of music ... often sparked a memory of events from the past*, while another observed that *well known songs promoted his memory, comforted him and made him feel fine*. A third professional noted that:

Music is one of her greatest interests, and she remembers several lyrics because she has sung them so many times ... the music helps with her memory.

A parent noted that listening to music helped the daughter to remember:

If she went to that particular concert, she would remember what the group were wearing or doing during a particular song. She will also remember what we did before the concert or any funny things that happened. This is really important for us as parents, as it helps not only with her communication but also her long-term memory.

Several parents mentioned that among young people with JNCL the songs from childhood seemed to be enjoyed and remembered.

Music was the most important activity in his life. He listened to music every day, played his drums and sang his favorite songs. His music taste changed later years towards children's songs.

Those supporting young people with JNCL can ensure that the music associated with pleasant and important memories can be retained by putting together "banks" of songs and other pieces, together with notes on when they were heard, and other details of the occasion, to help prompt recall (see Chapter 13). In this way, music can serve as a "bridge", as one parent put it, to a past reality, in the final stages of the disease.

Concluding comments

For individuals with severe visual impairment, hearing is the main compensating sense. Although most children and young people listen to music in many situations, the lack of vision can mean that music is even more important for children and young people with JNCL. There is, however, limited research on the educational use of music and song with this group, and it is not safe to assume that all the findings from research with other groups can be transferred to children and young people with JNCL. It should also be noted that research on music-based interventions for elderly people with dementia suggest positive effects of music in some areas, but results are mixed (Koger, Chapin, & Brotons, 1999; Raglio et al., 2015; van der Steen et al., 2018; Vink, Bruisnma. & Scholten, 2003). The present survey demonstrates the importance that parents and professionals attribute to music activities for children and young people with JNCL. This emphasizes the need for more research in this area.

References

- Baird, A., & Samson, S. (2015). Music and dementia. *Progress in Brain Research*, 217, 207–235.
- Bills, W., Johnston, L., Wilhelm, R., & Leslie, G. (1998). *Teach and be taught: A guide to teaching students with Batten disease*. Columbus, OH: Batten Disease Support and Research Association.
- Gylfason, G. B., Jóhannsdóttir, K. B., & Einarsdóttir, H. (2006). Living with Batten disease in Iceland: The story of Hrafnhildur. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 48–49). Örebro, Sweden: Swedish Institute for Special Needs Education.
- Heikkinen, I., & Railosvuo, S. (2006). Communication and computer assessment: The case of Jonna. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 50–51). Örebro, Sweden: Swedish Institute for Special Needs Education.
- Lewis, J. (2013). A cross-cultural perspective on the significance of music and dance to culture and society. In M. Arbib (Ed.), *Language, music, and the brain: A mysterious relationship* (pp. 45–66). Cambridge, MA: MIT Press.
- Morrongiello, B. A., & Roes, C. L. (1990). Children's memory for new songs: Integration or independent storage of words and tunes? *Journal of Experimental Child Psychology*, 50, 25–38.
- Ockelford, A. (1994). Music ... a source of enjoyment for all. *Information Exchange*, 40, 10–13.
- Ockelford, A. (1996). *All join in! A framework for making music with children and young people who are visually impaired and have learning difficulties*. London, UK: Royal National Institute for the Blind.
- Ockelford, A. (1998). *Music moves: Music in the education of children and young people who are visually impaired and have learning disabilities*, London, UK: Royal National Institute for the Blind.
- Ockelford, A. (2008). *Music for children and young people with complex needs*. Oxford, UK: Oxford University Press.
- Ockelford, A. (2012). *Applied musicology: Using zygonic theory to inform music psychology, education and therapy research*. New York, NY: Oxford University Press.
- Olsén, L., & Laine, S. (2006). Working with students who have Batten disease. In S. von Tetzchner (Ed.), *Proceedings of the First International Education Conference on Batten Disease* (pp. 72–73). Örebro, Sweden: Swedish Institute for Special Needs Education.
- Patel, A. (2007). *Music, language, and the brain*, New York, NY: Oxford University Press.
- Schön, D., Gordon, R., Campagne, A., Magne, C., Astésano, C., Anton, J. L., & Besson, M. (2010). Similar cerebral networks in language, music and song perception. *NeuroImage*, 51, 450–461.
- Serafine, M. L., Crowder, R. G., & Repp, B. (1984). Integration of melody and text in memory for songs. *Cognition*, 16, 285–303.
- Thaut, M. (1999). *Training manual for neurologic music therapy*. Fort Collins, CO: Center for Biomedical Research in Music, Colorado State University.
- van der Steen, J. T., Smaling, H. J., van der Wouden, J. C., Bruinsma, M. S., Scholten, R. J., & Vink, A. C. (2018). Music based therapeutic interventions for people with dementia. *Cochrane Database of Systematic Reviews*, Issue 7.
- Vink, A. C., Bruinsma, M. S., & Scholten, R. J. P.M. (2003). Music therapy for people with dementia. *Cochrane Database of Systematic Reviews*, Issue 4.
- Walkley, S., Siegel, D., & Dobrenis, K. (1995). GM2 ganglioside and pyramidal neuron dendritogenesis. *Neurochemical Research*, 20, 1287–1299.
- Woods, B., Spector, A., Jones, C., Orrell, M., & Davies, S. (2005). Reminiscence therapy for dementia. *Cochrane Database of Systematic Reviews*, 2, CD001120.

Musical Action to Support Children and Young People with Juvenile Neuronal Ceroid Lipofuscinosis

Jochen Lippe Holstein

As I enter the classroom, once again Marion sits completely limp in her wheelchair. School days have become very stressful for her, so she often lies down on the couch in the classroom. In the music room, I play her a new piece of music. The improvising soprano saxophone seems to remind Marion of the ducks at home, because she is particular happy about this sound. Together we dance to the music whilst standing. The dance, in which I let myself be completely guided by Marion, is getting wilder. She jumps and skips with all her strength, whereby she completely exhausts herself. We sit down again – now I give her a duck pipe which she uses to respond to the soprano saxophone. This dialog is also becoming increasingly fierce. To regain relaxation, we let the ducks fly away (becoming quieter as they fly away) and listen to quieter music whilst sitting, in which the saxophone also plays softly and slowly; we sway to the rhythm of music.

Marion loves music. My experiences with her are an example of the possibilities to support children, adolescents and adults with juvenile neuronal ceroid lipofuscinosis (JNCL) through «musical action». This chapter discusses how joint action within the music context with a degenerative disease may be an appropriate and meaningful support for development and maintenance.

Musical action

Human beings are characterized by having complex cognitive abilities. They are able to memorize experiences of the past, find relations and link them with other experiences. These experiences result from acts of purposeful activity and from managing the everyday world. Through these actions, human beings acquire knowledge about the world (Piaget, 1952), and the ability to take action is thus

the basis for the identification ability. Conversely, due to earlier experiences, it is possible to develop a differentiated ability to act. It is possible to observe the abilities of the individual and define goals that he or she may be able to achieve. These abilities are here divided into the domains of perception, movement, expression and communication. However, they are closely related, and influence and depend on each other (Amrhein, 1993).

Musical action means the arranging and the experiencing of musical contexts: playing, listening and responding to music. The everyday subject music is both a cause and an aim of perception, movement, expression and communication. Music addresses these four abilities in a special way, and it may become a common and wonderful subject for communication.

In this chapter, several aspects of musical experience are discussed. Some neurophysiological and psychological assumptions are elaborated. This distinction is purely for methodological reasons, because music is always experienced individually *and* in a context – «music as a whole».

Music as sensory-motor experience

Music is not solely an acoustic phenomenon: perceptual information arrives through the auditory and tactile senses of the experiencer. For example, the stimulation obtained from an orchestra playing is experienced simultaneously through the sensory cells («hair cells») in the cochlea, within the ear, and through sensory receptors on the skin activated by the air pressure from sound waves generated (vibration) from musical instruments. Music can therefore be both felt and heard. Someone playing music also receives additional haptic information from the musical instrument itself (e.g., holding a mouthpiece against the lips, or plucking or bowing strings).

From a neurophysiological perspective, perception is not limited to the sensory afferent of environmental stimuli. An efferent motor order is often the result, which is carried out either deliberately or involuntarily, as a reflex or automatic response. It appears to be a close link between sensation and motor activity (see Gibson, 1979; Liberman and Mattingly, 1985; Guenther & Vladusich, 2012). In addition to the heteroceptive perception (on the outside), the human organism also has a proprioceptive perception, a sense related to self-awareness, such as balance or sense of movement (the vestibular and tactile-kinesthetic sense). Heteroceptive hearing and proprioceptive self-awareness are related structurally and functionally through anatomical proximity.

Whilst music motivates movement, it moves humans in two ways: The person feels the tactile-kinesthetic vibrations and the movements of his body parts, the vestibular and the spatial variation in his body, and is moved emotionally!

Moreover, a noise or sound can only be produced with movements. Thus, to make music, targeted and coordinated movement is necessary. Music speaks to the human organism in a diverse sensory and motor activity interrelated manner, very unlike other contexts of actions or action objects. Music is perceived as sensory-motor activity in action.

Music as personality of action

With music, I here mean music that originates from humans and where material is consciously brought to make the sounds which are used for the subjective view or subjective expression. In each case, the phenomenon of music is malleable, since it is produced and perceived by humans and thereby structured. Emotional content is perceived as sensory-motor as well as cognitively detectable structures, object-like features can be illustrated, associations are activated, music challenges the person to make a statement. When the individual has visual impairment, differentiated acoustic perception provides important information about the environment. The distinction of objects as sound sources is also facilitated by the recognition of (cognitive detectable) structures, which also facilitates spatial orientation. The sound quality provides further information about the nature of the sound-making object. These qualities can be used consciously to express something specific in a music-making process. The malleability of music gives the person opportunity to express himself through musical activity, to express emotions, illustrate thoughts and thereby visualize and process. An emotional expression can be put into a piece of music, or it can be experienced subjectively within a piece of music.

Music as an independent form of communication

Musical action can be understood as a communicative process, because between the music interaction partners, defined and definable symbols are purposefully replaced, similar to language.

However, a special feature of musical communication is that there exists a possibility for the person to develop his own symbols, or even intersubjectively to define symbols with instrumental or vocal sounds. As a result, a relationship can be established between the music performers, something can be expressed or described through music (Reinecke, 1982).

In contrast to verbal communication, the focus is often a more idiosyncratic expression with musical and non-verbal communication: There is no right or wrong information, although culture-bound evaluation criteria may be created in regard to embodiment and performance shape. In musical action, communication can take place on a very basic level. For example the music therapist may take over

the rhythm of the other's breathing, start to make contact and thereby put forth an equal level. The music therapist may change the rhythm playfully, insert a new aspect in the relationship, to which the partner can consciously respond (stay at his own pace or acclimatize himself to the new rhythm). Musical dialogues can be highly differentially designed, if both players adjust to each other, respect common rules and can use differentiated skills and abilities. In any case, through musical action the participants create call-and-response sequences in which an action (e.g., an utterance or rhythm) of one participant evokes a related reaction (e.g., imitation of variation of that utterance or rhythm) from another participant. If both participants refer to the same rules, musical action can result in an expectable communication – or a surprise is deliberately presented by consciously violating the expectations. In any case, both people are actively involved in designing a communicative action.

Unlike linguistic communication, a simultaneous polyphony can even be desirable and consciously used to express something condensed or parallel. Musical communication therefore has usages other than verbal communication. It can approximate and mimic phonemes – music can however deliberately use the differences to supplement the verbal action.

In a group context, musical action provides a very diverse communication frame for the fellow players. They can focus their attention, perception and movement on a common content, in order to play, listen and experience music, and coordinate their movements in such a way that a uniform process can arise. It can be a wonderful experience when a whole group of people is so closely related and involved in the same process!

Nurture despite degenerative disease

The aim of educational activity is to support in the individual a sense of differentiation of skills and abilities. Strictly speaking, this means the differentiation of each ability in action, that is, the ability to act purposefully in a specific context. Jetter (1987) defines the ability to act as «The ability in the given situation and for the anticipated living conditions, the greatest possible solidary use of natural and social conditions for the purpose of a multiplicity of experiences indulgency, according to the physical and mental needs and the ability to aesthetic-communicative shaping of the life situation» (p. 79). Jetter refers to the work of Piaget (1952), who described development as a dynamic connection between the individual and the environment as follows: Every human organism strives to live in harmony with its environment. The young child forms structures of action schemes, in order to adapt to the surrounding conditions (accommodation),

and then again in order to assimilate the environment to his own structures (assimilation). In the young child, these two adaptation processes run as simple activities of the actual need of satisfaction. With further development, conscious, planned action shapes the more complex connections between the individual and the environment. The changing conditions of the environment (in Jetter, these are the «natural and social conditions») are perceived and taken into account. Likewise, the structures of the developing individual change continuously. When a child or young person has JNCL, the structural change involves a reduction or loss of already acquired possibilities for action (see also Chapter 2). The degenerative disease is thus to be understood as a component of the «given and expected living conditions» (see Jetter, 1987) of persons with JNCL. Their change is also development, as adjustments need to be made, but they also need support. By motivating pupils with JNCL in their subjective and actual reality – that is, their ever-changing capacity for action – the teacher encourages purposeful activity.

Music as an occasion and goal for common action

As explained above, growth is achieved by appropriately addressing the abilities of the individual within a context, which he recognizes as valuable. Music presents itself for shared action, since it can be experienced in different and diverse ways. This ensures that musical action, even under changed and further changing conditions, in particular with the limitation and loss of abilities, is a valuable mind context over a long period of time. Between the educator and the student, a private music or a private musical understanding can arise. The structures can only be recognized and understood by them, as they reflect their personal relationship. Changes in the musical capacity for action therefore do not have any hindering effect on the quality of the mutual musical life. For example, in spite of limited motor possibilities in the mutual sense of the time, music can be played together, interdependently (see Chapter 16). The pupil may play only every fourth note while the teacher continues to play the complete piece, but they play together and the pupil is actively involved and experiences himself as effective and with equal entitlement.

If the orientation to objective assessment criteria within a musical context (playing the piano correctly) represents an excessive demand or a motivation obstacle (especially when playing well-known pieces of music), the rich range of musical trends and styles can be relied upon at any time, allowing each person to choose the music which appeals to him. In musical improvisation, for example, subjective assessment criteria can be implemented without the impression that the result is a makeshift product or something provisional. Musical improvisation

is therefore particularly appropriate, because subjective changes in the game that result from the disease-induced change in the ability to act can be sensibly absorbed.

Promoting the general capacity to act through musical action

Humans receive knowledge about the world through the active experiencing of parts of this world. In this case, music is such a part, which can be experienced through action: «In acting [that is, through the purposeful use of the capacity for movement, expression, perception, and communication], the subject reveals that the ‘outer’ reality music is accessible and understandable, and so on. At the same time, the inner ‘subjective musical reality’ – the ability to move, express, perceive and communicate – is developed» (Amrhein, 1993, p. 571). With regard to Jetter’s (1987) definition of ability to act, music can be regarded as a «natural and social condition», because it is experienced as an acoustic and tactile phenomenon, as a component of nature, and, on the other hand, as an eminently shaped component of culture. The notion of «experience of multiplicity» as the purpose of capacity for action has already been discussed. Jetter emphasizes the physical and spiritual need of this experience and that the «ability to aesthetically and communicatively shaping of the life situation» is evidently promoted through musical action.

Positive influence of the psychological experience through musical action

The damage to the nervous system in JNCL (see Chapter 3) does not lead only to impairment in motor and cognitive domains. Children and adolescents with JNCL also notice accompanying changes that occur. For example, they experience multiple losses, such as the loss of already learned or acquired abilities and mastery of activities, as well as loss of social contacts and ties. They also experience the changing responses from the environment towards them, such as disillusion, worry, anxiety, anger, perplexity, and constant observation and classification. Since they recognize the cause of these changes in themselves, they also often feel responsible for the reactions of their surroundings. Moreover, they become massively unsettled when information about their future is lacking or unclear, and fears arise.

It is often observed that taboo subjects, such as illness and death, are avoided or not dealt with by everyone involved – the young persons with JNCL as well as significant people in their environment. Since there is never the opportunity to talk about such serious topics in everyday life, young people’s concerns (e.g.,

fears, anxiety, uncertainties, misunderstandings) arising from these topics always remain diffuse and are never openly addressed. If at a later stage, the ability to speak or use language are lost, this situation is further intensified, because the young people's unaddressed concerns remain and are compounded because it is now more difficult to address them openly.

Therefore, it is necessary to create a framework within which crisis topics can be dealt with. Conditions for this are a trusting relationship and an appropriate medium of expression. Musical action allows an active symbolic examination of one's own thoughts and fears, especially when the medium of language is increasingly impaired. The aim is to address such topics and prevent or take away the paralyzing hopelessness and menace, by showing confidence and confirmation in the present moment, which can contribute to a positive evaluation of the young person's identity and development.

How to make music together

First of all, I would like to emphasize that every offer of action must be in a context that is meaningful to the pupil and within the pupil's subjective reality, so that he or she can use their current competences. Depending upon the severity and extent of the disorder, the body and its tools can be recruited to make sound, for example in rhythmic breathing or knocking games (anticipations and reactions in the form of imitation or variation), or in imitation of noises or operations, thus allowing for whole sound stories to occur. When participants are singing songs, they can invent sensory or rhythm-supporting movements.

The voice is the person's most obvious medium of expression, and it is usually used for speaking and singing. In speech, the content is relayed by means of words which are intended to represent what is rationally detectable in a conventionalized manner (e.g., in statements, questions, or commands). However, the spoken statement can be modified by paralinguistic means (prosody, patterns of intonation, modulation of loudness and pitch), and non-linguistic means of expression, (e.g., kinesics including gesture and facial expression; proxemics, such as spatial closeness; and chronemics, including the relative timing of communication). When meaning is conveyed through the medium of singing, the non-verbal aspects employed are of special importance. The sounds of the voice (i.e., patterns of intonation, modulation of loudness and pitch) can be used to express, for example the personal significance of events, objects and people, by using them phonetically. Conventional and homemade instruments lend themselves to taking musical actions, such as exploration and experimentation. It is also possible for participants to perform the same reaction and imitation games as in the breathing

and knocking games, and also to create a rhythmic accompaniment of songs or other music pieces. For this purpose either drums or other rhythm instruments or (bass) tones are useful for harmonic-rhythmic song accompaniment.

Listening to music motivates music-related movements. These movements can be used to represent parameters of the musical process – including structure, dynamics, tempo and pauses. The vibrations of large gongs or basins or of bass instruments are perceived as pleasant and relaxing, if they are embedded in that kind of context, for example telling a story or fairy tale. The perception of these vibrations usually stimulates responsive movements. These movements are experienced as a meaningful activity within this situation. It is quite pleasant and meaningful to just listen to music. This also gives the opportunity to be «completely within the music» or «completely by oneself.»

Examples of musical action

The next section presents situations of shared musical action with two young people who have JNCL, «Klaus» and «Marion». Their current competences, the related goals and the teaching situations are described and discussed.

Klaus

Klaus is 14 years old and attends a class in the department for multiple or severely disabled children in the National Education Center for the Blind. He is a very lively, daring and communicative boy. By now, most of the time, Klaus makes a well-balanced impression since he feels very comfortable in his class. Some days he is rather obstinate and more confrontational than usual. When I first met Klaus some years ago, I noticed that he was tense and displayed a certain aggressiveness. I put this down to the fact that he could not accept his new situation. He had recently been moved from the mainstream part of the school to the part for people with multiple disabilities. In his new class, he had little opportunity to communicate his knowledge or even to compete with other pupils, since there were no equal conversational partners among the other pupils in the class. A constant, self-produced performance pressure could not be satisfied. After a further change of class to the current stage of the lower classes, more favorable conditions were created for Klaus. He was able to establish appropriate contacts with peers who were at a comparable intellectual and emotional level.

Klaus is inquisitive, but he also likes to share his knowledge with others. For every topic addressed, Klaus first tells me what he knows, and it is often quite extensive. Klaus knows something about everything, and he can make connections

from anything to topics that are familiar to him. However, like many adolescents with JNCL, Klaus has significant difficulties with speech. He talks very fast and blustering, deletes or reduces words and syllables, repeats himself frequently, and sometimes does not find the right words. Klaus can orientate himself and can generally walk without any external support. However, when he walks, a knee flexion is noticeable, which may be observed in most young people with JNCL. Occasionally Klaus has a problem keeping himself on his feet. Increasing knee flexion when he walks appears to compromise his balance. In many situations, his muscle tone is increased and his movements appear tense. He often sits with raised shoulders and crossed legs, and his hands clasped under his thighs. To demonstrate power and compare himself to others are important to him. Klaus has good fine-motor skills and is motivated to use his hands.

Support goals

Helping Klaus preserve his self-image is a priority when one is working with him. To do this one must ask numerous questions: How does Klaus see himself in his development? What goals or ideals does he pursue and why? Why does Klaus claim that he can see, although this is actually not true? One goal consistent with preservation of self-image might be to look for possibilities of physical and psychological relaxation for him, since in the long term both will be of increasing importance. In order to be able to adequately counter possible instances of restlessness, it is of great benefit to know the individual relaxation preferences or framework conditions.

In addition to the goals mentioned above, which primarily aim at his mental well-being, is a goal to support him in maintaining a certain range of movements. Movement can be for fun or as a means of pursuing a goal. However, maintaining gross and fine-motor functionality, whether for mobility or participation in activities has special relevance to the later need for care.

The individual sessions with Klaus are designed in such a way that either a range of materials or the general framework of the treatment remains the same for an extended period of time. The pre-setting of action topics or action material initially opens up a common scope, which can then be designed individually.

A story as an occasion for musical movement action

Reading a story aloud functions as an opener for presenting topics and tasks for individual sessions. Today, a story that stretched over half a year ends with a festival of sports activities. «The butterfly workshop» by Gioconda Belli (1994) is about the origin of many species, and to celebrate the creation of the butterflies, a feast is arranged with self-invented program themes, including «Dance of the Butterflies», «Tug of the Spiders», «Boxing Match of the Kangaroos» and «Soccer

Game of the Moles.» Activities included in the last two program themes are described below.

In «Boxing Match of the Kangaroos,» a boxing match is symbolically represented by beating the fists or using mallets on a timpani (kettledrum). Klaus and I are sitting opposite each other. At first Klaus plays (punches) with his hands: quietly and slowly or loudly and fast, and then I change strategically, mimicking him. The roles of being initiator and responder change equally. The relationship to a real boxing match and, above all, the idea of kangaroos boxing, has a motivating and stimulating effect. Klaus tries out different techniques – on the drum ring (box ring) and on his own chin, in order to experience the effect of a drum beat.

«Football game of the moles» is played in a self-made carton football field table. We both try to flip or shoot a marble with our hands in the opponent's goal. The game is characterized by rapid changes between fast and quiet phases. Klaus locates the sphere partially by using his hearing, partially with searching hand movements. The «football lingo» is of great importance to Klaus. The relevant accompanying comments, shouts, and so forth, recreate a real football game.

Search for expression

One day, before the start of an individual session, I was sitting at the piano and improvised a little before Klaus came to the music room. He asked me if I could play something sad. I did. Klaus, at that time very restless and hyper in his mood, listened quietly at first, then he told me about deaths in his circle of relatives and acquaintances. He asked what happens after death, and how or what death is at all. Then I was reading some sections from the «Letter to a child with cancer» by Elisabeth Kübler-Ross (1979). The illustrative examples (seasons, day and night of Earth) seem to be understandable to Klaus. He questioned the examples and showed thereby that they made sense to him. Klaus himself does not use music to express sadness; for him, making music is something powerful and joyful. However, he listens to melancholic music and therein finds the desired expression of his feelings.

Marion

Marion is 16 years old and goes to middle school. She attends school irregularly because of frequent seizures and a growing demand for rest. Marion has long been dependent on the wheelchair for mobility. With maximum support she can walk small stretches or dance or rock to music, which she likes very much. Her posture is JNCL-typical, with forward inclination. Without a backrest or other support, she does not have sufficient support for sitting, and sitting unaided is hardly possible. Marion is stiff in the joints and each movement is met by counter-

pressure. Activities are often exertive, even lifting her head requires a great deal of effort. Therefore, after most movements she returns quickly to her flexed posture. In her classroom is a sofa on which she spends more and more of her time, as sitting a long time has become very demanding. Marion frequently appears tired or absent-minded.

Marion's comprehension of words has developed in accordance with her age. She understands every message and request, even ironic remarks. Based on her reactions, it is evident that she follows the topics and forms her own opinions. However, her expressive language is less functional. Marion speaks very quietly and gently, and seems to lack the air to speak. It happens often that the air is not sufficient for her to reach the end of a sentence, so that the last words are incomprehensibly weak or not pronounced.

Although Marion is very quiet and reserved, and hardly able to express herself verbally, it is apparent that she enjoys being with other people and experiencing things that interest her. When asked if she wants to do this or that, she often reacts with a shrug, but sometimes she very enthusiastically agrees to a suggested activity. Especially when she is part of a group and involved, her joy can be observed. Marion likes to accept certain offers. Likewise, she is very fond of being «part of the scene,» she is happy when many people around her do something that she can be part of, usually meaning listen to.

Support goals

In the work with Marion, the focus is her ability to move, in order to maintain her expressiveness and scope for action. Through the active participation in situations that are meaningful and interesting to her, many movements and forms of communication may become activated. Since Marion cannot express all her ideas in speech, debates in the form of conversations are not appropriate for her situation. Under certain circumstances, topics which she would like to address may be torn, because of her restricted ability to speak, and this could lead to frustration. That is why the sessions are used to develop our own communication symbols, by means of which an inter-subjective expressive possibility is created.

Musical participation history

One day, I had prepared a participation story for Marion. The content of the story was built on Marion's interests and preferences, so that she could make a connection to the storyline. Through the following activities, she was given participation opportunities for action: different animal sounds were produced, the story ended with a joint celebration of animals and dwarves, which Marion and I use for dancing. Marion enjoys dancing very much, she is able to stand when support is provided.

The individual sequences – how the animals are represented with sound, how the animals react to certain events – are made so that Marion can take part independently. Marion replies to me according to her possibilities in the form of imitation or variation. Of particular importance for Marion is the representation of the ducks. Her parents are breeding ducks at home, which is why Marion has an exact idea of how these animals sound for example when they are excited. We use a duckling pipe to produce the typical quack-cackle sound. However, it contributes only a sound representation: Marion represents a duck with the whole body. She expresses «duck» on the basis of many duck experiences. Marion, who usually is very quiet, speaks gently and sits in a bent position, raises herself to fill her lungs so as to give the ducks a voice.

Improvisation around annoyance

When I picked up Marion from her classroom one day, she was sitting listlessly in her wheelchair, punching without interruption – but also without power – on a pillow filled with sand. She reacted to my greeting, but continued with the treatment of the pillow. In the music room, I asked her if she is annoyed. She answered «yes.» In the further conversation, I did not find out what had made her so annoyed, but we had chosen «annoyance» as the topic of the session. The sand sack was treated in a variety of ways, such as stroking it like a beloved guinea pig, and beating it like it was something dangerous, and soon drums were drilled. In the meantime, Marion had freed herself up by playing. She sat upright in the wheelchair, with raised head and a concentrated and content facial expression, enjoying the volume that she caused. We took turns bashing and stroking, and soon the drumming developed into a musical act. We took turns with drum whirls and signals – we made music together.

Conclusions

Musical action is a pedagogical principle that provides suggestions for the education of children and young people with JNCL. The stories presented here illustrate the ways in which pupils' interest areas and abilities may be used to establish joint musical action. The beauty of the music, and the use of its formality are clearly visible in the representation of the animals and their activities in the stories. However, even in search of expression and in improvisation, the capacity for personal expression, including the expression of annoyance, was only possible because it could be found in the music elements and transferred into the children's own musical actions. What the two pupils really felt was malleable and can only be observed by an outsider by looking at their actions. The fact remains however, that

the two pupils could find expressions of sadness or annoyance in the music. Marion expresses her anger through these musical structures. The exchange between the pupils and me did not only take place at the vocal verbal level. In the boxing match, we spoke mostly with music accompaniment, after that, the game was reversed, thus accompanying music-playing. Musical parameters (e.g., dynamics or tempo) determined the course of the action, not grammatical, syntactic or other linguistic parameters. Likewise, a chaotic and two-part drumming session represented Marion's improvisation of anger excellently, perhaps even more appropriately, with greater social acceptability than would be possible to express through language. The sensorimotor experience of music in musical action is always recognizable. This is most evident in scenes where dynamics and tempo are experienced physically as tension and relaxation, such as in boxing or experiencing the duck. However, even in the quiet football game, the rolling of the marble was sensory-motor experience.

In musical action there exist possibilities for targeting therapeutic goals by exploiting earlier experiences that the child recognizes. This therapeutic strategy is motivated by the observation that pupils perceive the musical action in itself and recognize its value. The encouraging – and probably also motivating – common factor in musical action is that different people orient themselves towards a goal, influence each other in their actions, and thereby enrich each other.

A final aspect of musical action of great importance is that musical experience is possible even when the neurodegenerative process reduces motor action possibilities!

Note

This is a revised version of an article in «JNCL. The living situation of blind children and adolescents with an incurable degenerative disease» from 2001.

References

- Amrhein, F. (1993). Bewegungs-, Ausdrucks-, Wahrnehmungs- und Kommunikationsförderung mit Musik. *Zeitschrift für Heilpädagogik*, 9, 570–587.
- Beierlein, J., & Held, J. (1986). Förderung Schwerstbehinderter mit Musik. In K. Hörmann (Ed.), *Musik im Diskurs I. Musik- und Kunsttherapie*. Regensburg, Germany: Gustav Bosse Verlag.
- Guenther, F. H., & Vladusich, T. (2012). A neural theory of speech acquisition and production. *Journal of Neurolinguistics*, 25, 408–422.
- Hegi, F. (1993). *Improvisation und Musiktherapie*. Paderborn: Junferman.
- Jetter, K. (1987). Erziehungswissenschaftliche Grundannahmen einer handlungsorientierten Didaktik. In F. Schönberger (Ed.), *Kooperative Didaktik* (pp. 78–82). Stadthagen: Bernhardt-Pätzold.

- Kübler-Ross, E. (1979). *A letter to a child with cancer*. Escondido, CS: Shanti Nilaya.
- Liberman, A. M., & Mattingly, I. G. (1985). The motor theory of speech perception revised. *Cognition*, 21, 1–36.
- Piaget, J. (1952). *The origin of intelligence in the child*. London, UK: Routledge and Kegan Paul.
- Reinecke, H.-P. (1975). Kommunikative Musikpsychologie. In G. Harrer & C. G. Allesch (Eds.), *Grundlagen der Musiktherapie und Musikpsychologie* (pp. 111–122). Stuttgart: Fischer.

Technology in Education

Mohammed Beghdadi

Technological advances have resulted in an extended use of technology in many areas, including education (Craft, 2012) and rehabilitation (Cook & Polgar, 2008; Scherer & Glueckauf, 2005). Technology is used to support and maintain sensory functioning (Edwards, 2007), motor skills (Henderson, Skelton, & Rosenbaum, 2008), and cognitive functioning (Carey, Friedman, & Bryen, 2005) in children as well as older people. The aim of introducing technology in special education is to adapt the teaching to the needs of each student. Adapted teaching does not necessarily imply a particular way of teaching, but rather a situation that is characterized by certain qualities that make students and teachers develop together. The teachers put themselves in the students' situation in order to acquire information that will improve the classroom situation for students with special needs (Buli-Holmberg & Ekeberg, 2008). The development of the students' competence, coping, self-determination, co-operation and appropriate work habits is crucial for successful learning.

The present chapter gives an overview of assistive devices and tools that may be useful in teaching students with juvenile neuronal ceroid lipofuscinosis (JNCL). The devices and educational tools presented here may be useful for students with JNCL but most of them are not designed with this group in mind. Children and adolescents with JNCL have generally had very limited access to computers and teaching aids that are tailored to their particular needs. The combination of visual, motor, language, and cognitive impairments and challenges in social interaction makes it difficult to find an appropriate way to cater for their needs. There is a need for more devices and tools designed for students with JNCL, and these may also be useful for other people with disabilities.

The tools presented here have two basic purposes: to compensate for declines and other educational needs and to support participation. Tools, including electronic braille displays (see Chapter 14) and devices with voice recognition and production, can serve to compensate for declines in visual, cognitive, language

and motor functions. The compensation thus provided can in turn provide these students with support in participating in educational and other activities at school, at home and with peers outside school, thereby stimulating them to maintain previous patterns of participation, and possibly encouraging them to maintain and extend those patterns even in the presence of decline. The use of technological tools in education may enhance the students' confidence and self-esteem, and function as help to better inclusion through independence and interdependence (see Chapter 16), and thereby to increasing the students' benefit of the teaching. The use of technology may also contribute to increase motivation, engagement and achievement efforts in areas where the students need support, training and practice. It is the experience of the present author that the use of technology and innovation in education may contribute positively to an interesting school day for the students with JNCL and give people in their environment a better understanding of the students' opportunities and abilities, for example when the student is using the specialized computer program Sarepta (see below). Technology is also used in games (see Chapter 21) and quizzes (see Chapter 20). Some of these technologies may also have a positive impact on adult life after finishing school.

Aims, possibilities and challenges in using technology

One of the main aims of using assistive technology for persons with severe visual impairment, including JNCL, is to make information available through touch and hearing (Freitas & Kouroupetroglou, 2008; Petty & Frieden, 2012). Assistive technology programs can be used to translate spoken language into digitized text, turn spoken commands into actions or magnify the text in a word processor, web browser, email programs or other applications. Assistive technology has contributed to removing many barriers for persons with disabilities within education and employment. Possibilities and goals within these areas are also valid for persons with JNCL. Students with JNCL can for instance complete their homework, do research, take tests, and read books along with their sighted classmates, thanks to advances in technology.

Assistive technology tailored for people who are blind makes it possible to organize and handle files and records containing text, music, audio-recorded memories or almost anything that can be stored in the electronic world. Assistive technology can also be used to support inclusion. Students with JNCL can for instance share the same classroom activities as their peers by using technology where visual information is transferred to sound or tactile information (see Chapter 14). Further, some programs and devices may be used in some situations

to compensate for loss of speech, memory and mobility. Last but not least, use of assistive technology can provide entertainment and pleasure at all ages for persons with JNCL within and beyond education (see Chapter 23).

Assistive technology for persons with special needs is often associated with possibilities for independence. This is a mandatory goal also for children with JNCL when independence is still achievable. Independent use of assistive technology will however require understanding and skills related to the operation of electronic devices, and this will require a certain degree of memory and motor functions.

Technology-based information can be operated simultaneously or sequentially. Visual information allows simultaneous actions where segments are presented in a two- or three-dimensional hierarchy or structure. The user with sight can select an option by ticking the desired segment presented in the structure, a glance on the structure is enough to identify the correct option. To transform such structures to tactile or audio-based information requires a more complex strategy. First, pictures on a screen can only be transferred as time-consuming narratives. Second, tactile and audio-based information requires sequentially-based or consecutive actions where the user follows a string or succession to identify the correct option. This step-by-step strategy requires time and memory: the user must remember the purpose of the string, and previous actions to enhance the next actions. Sequentially based



Figure 19.1 Rolltalk Designer

Rolltalk Designer is a tool for communication programs for persons with disabilities. The program has a large amount of finished content, both symbolic and textual.

information in assistive technologies is a special challenge for persons with JNCL when memory problems are evident, a finding emerging from the experiences recorded in the current project. This memory demand might be a main reason why electronic communication aids (see Figure 19.1) are not used to a higher extent by persons with JNCL. In addition, there are significant demands on attention because someone using the technology must be able to filter out background noise, remain focused on the task and so forth (see Chapter 6).

Assistive technology is used in a range of areas and for a variety of purposes, such as performing daily tasks and activities, supporting social participation, monitoring health and improving safety; however it is not without limitations: Assistive devices can never replace human contact and interaction for persons with JNCL. Skills and functions will decline for individuals with JNCL (see Chapters 3–7) and independence will sooner or later not be obtainable. When that point is reached, *independent* use gives way to *interdependent* use, allowing assistive technology to continue to have an important and positive impact on life for persons with JNCL even when independence is difficult to achieve. Objectives related to sharing memories and pleasure, and compensation for loss of speech and other functions can be achieved by using interdependence strategies where assistive technology is used with assistance (see Chapters 16 and 23). The long-term goal of using assistive technology for persons with JNCL should consequently be less about its facilitation of independence, and more about its supportive role for facilitating participation and interaction through interdependence strategies.

Compensation through use of audio devices in teaching

Assistive technology can support functioning in the zone of developmental maintenance (see Chapter 2 and 11). As the disease progresses, children and young people with JNCL can compensate loss of vision with information received from touch and hearing. The present project (see Appendix A) found that the tactile sense tended to weaken earlier than the auditory sense (see Chapters 1 and 14). It is therefore important to consider assistive technology and teaching methods based on hearing.

The use of audio and video media in education has grown considerably in recent years, although not always with success. Constraining factors are lack of technical standards, technological expertise in schools, and technological and educational guidance and supervision (Craft, 2012). However, for students with visual impairments in general and students with JNCL in particular, the use of audio provides opportunities for improved learning and participation.

In early phases of JNCL, the use of audio for presenting texts should usually function as a supplement rather than as a substitute for reading and writing (see Chapter 14). Reading and writing are pivotal skills for further learning and development. In the present study (see Appendix A), some students with JNCL had learned to read and write braille or Moon, others only managed to write in the tactile mode, while some never managed to utilize tactile texts. With the support of a speech synthesizer this last group could nevertheless have access to written texts through listening. Moreover, hearing will sooner and later become the most important sense for participation and learning in all individuals with JNCL, and assistive technologies with speech synthesizers should therefore be considered as soon as possible after they have received the diagnosis.

The present study shows the importance of making sound recordings of early situations of special importance for the student, such as conversations, interviews, stories, and excursions at school, as well as presentations, jokes and expressions that the students used in different situations. Audio recordings can be used to preserve memories for the future, as in the following narrative from a teacher:

The student had used the program "Sarepta" for several years. As part of the pedagogy, the staff at her school had documented the student's school day by recording a lot of activities and school assignments. For many years, the parents recorded family trips, travels, birthdays and special adolescent events like the Christian confirmation on video and audio. She had a unique, extensive and comprehensive audio bank from her childhood and adolescence years, with material she produced at school, such as interviews, music and dance videos, workshops and school graduation celebration. When the student became an adult and her illness had reached the last stage, listening to these recordings was one of the few activities she could take part in. She responded to what was being said, expressed emotions and sang along to her old childhood songs. The recordings had now become a medium and a tool for communicating with her closest family and friends. The mother stated that this was the best activity they had together, where they could communicate and reminisce.

Audio can function as an inclusive medium. Collective listening to sport, radio plays, audio books, poems or music gives students opportunities for sharing experiences with others. However, it is important to be selective in what to save for the future, and how audio recordings are saved and stored for the future, how the files are edited, labeled and categorized. An overwhelming number of audio recordings, which are not labeled or categorized in a manageable way, will prevent future use of important recordings.

Compensation through use of speech recognition

Mainstream technologies such as Voice Assistants (for example Apple Siri, Microsoft Cortana, Google Assistant, and Amazon Echo) are today used in the mainstream classrooms and many other everyday settings (Jana, 2009; Shanmugapriya, Gayathri, & Deepa, 2016). Speech recognition technologies, speech-to-text, can recognize spoken words. The user talks into a microphone and words are transformed into a text file. Some of these technologies need to be *trained* to recognize a specific voice (Anusuya & Katti, 2009).

Speech recognition and speech-to-text programs have a number of applications for users with and without disabilities. These technologies can promote independence and autonomy and improve the individual's participation in the society (Shadiev, Hwang, Chen, & Huang, 2014; Silver-Pacuilla & Fleischman, 2006). The present study shows that some individuals with JNCL are benefiting from using these technologies, for instance when accessing and commanding the devices to do different things. Speech recognition requires however a minimum level of intelligibility and fluency in speech and cannot be used by persons with comprehensive speech problems. The usefulness of speech recognition for some individuals with JNCL should not be underestimated, as the following story illustrates:

A young man with JNCL was blind and used several electronic devices. He used speech recognition (Apple Siri) to perform daily tasks which he had not been able to do earlier. He was an active user of a website where he shared thoughts about music and activities. He managed voice calls and messages independently by using speech recognition and making commands like «Call James» or – «Send a message to Yvonne, I will visit you this evening». The young man was also able to store reminders, including date, time and activity. The young man found information on Internet using speech recognition, he simply asked questions like «What is the capital of Germany?» Speech recognition made it possible for the young man to access emails, calculators, alarm clocks and games, to make notes and to do his favorite activity – listen to music.

Assistive devices to support inclusion

How successfully students with JNCL prosper and function at school will largely depend on the social environment and the relationships with other students (see Chapter 22). The use of assistive devices can play a significant role in supporting

peer relations in the class. Some assistive devices are advanced multimedia tools for children and adolescents with communication difficulties. Many computer programs and devices may be regarded as socio-educational tools that give students new social opportunities, especially programs with a low entry level, such as social media, games, chats and music. Comments from parents and staff in the present study also show that many students with JNCL had become ardent users of Facebook and YouTube. Some assistive devices may be used for maintaining interests (and skills) and social participation when functions decline. The following story is about a young adult with JNCL who was a gifted piano virtuoso:

Using the music program Magix, a young adult with JNCL put self-produced music videos on YouTube. He was the acting composer and the music was produced and put on YouTube with assistance applying the Team Model (see Chapter 23). The person himself, his parents and friends, and general users of YouTube had great pleasure in watching and listening to the music videos. The composer with JNCL has received many remarkable feedbacks and comments on his productions from far and near.

Assistive devices to promote motivation

Student motivation is of great concern in education. Some students continue to work on an activity or assignment despite great difficulties, whereas others give up very quickly. Students' motivation is a main driving force in all learning activities and it also supports their self-efficacy (Bandura, 1997, 2006). Students' motivation influences their academic achievements, and what they learn and achieve will in turn influence their motivation. «In other words, it is not the actual learning situation itself that is central, but the students' own apprehension of themselves as learners» (Sølvberg, 2003, p. 17).

Some programs and applications presented in this chapter may function as motivation-generating activities for persons with JNCL and help them to accomplish many everyday tasks. Some programs have been developed in collaboration with the parents, school staff and housing staff with the intention to support work on school subjects and strengthen the students' confidence and self-esteem through achievement.

Opportunities for repetitions is a basic strategy to consolidate learning, in particular in individuals with memory problems (see Chapter 5; Cull, 2000). Quiz games may be used for achieving over-learning, based on necessary repetitions of

content, alternative answers and responses. Comments from parents and staff in the present project indicate that Quiz games in addition were perceived as fun by many of the students with JNCL, and quizzes were used to support motivation, learning and inclusion with peers (see Chapter 20).

Some tools that may be useful in education of students with JNCL

Although technology can provide enrichment to the learning process, deploying technology does not necessarily lead to better learning for anyone, regardless of whether they have a disability or not. Success will depend on a match between the technology and the user, the purpose of the technology in accomplishing a learning objective, and the choice of the most appropriate tools for the task (Cook & Polgar, 2008). Nevertheless, technology can help make learning interesting and it might be essential in making learning accessible. There is still a lack of educational software adapted for students with disabilities, but there is a growing range of mainstream technology, both hardware and software, that can serve useful functions for people with diverse characteristics and needs. In the following sections, a diverse range of tools will be presented. Some tools can serve many functions, and it is therefore difficult to categorize these tools. However, tools that are important to the student should be included in the student's curricula and IEP (see Chapter 11).

Many special utility programs for use by teachers have been developed. These programs enable teachers to create applications that suit a particular student's needs. Examples of such applications are Rolltalk Designer (see Figure 19.1; <https://www.abilia.com>), Grid (<https://thinksmartbox.com>), Book Creator (<https://app.bookcreator.com/>), Tobii Communicator (<http://www.tobiidynavox.no/>), Clicker (<https://www.cricksoft.com/uk>), and Sarepta (<https://www.regionorebrolan.se/sprida>).

Different software programs and apps may require different computer skills. Findings from the present study (Appendix A) suggest that some are easy to use while other programs require technical knowledge and practice. These applications often give instant feedback on answers, and some programs can be adapted to the student's level of functioning.

Tools for supporting communication

Students with severe expressive communication impairments have difficulty communicating with peers and adults and may benefit from using assistive technology to support communication. Many factors may contribute to communication difficulties: a decrease in speech intelligibility from motor dysfunction, a decrease in ability to put one's ideas into words, and decreased memory for topics one might want to share. The tools listed below can contribute in very different ways to providing support for communication. Thus, it is important to consider each tool from the perspective of how it might help a given student communicate with others. One must reflect on the communicative demands and the challenges faced by both the communicator and the specific communication partners.

A range of low- to high-technology solutions are available, including communication displays with objects, communication boards and books, switches with speech output, communication devices with synthetic speech and computer programs for communication (see Chapter 13). There are communication devices in the form of software, apps or analogue tools. Programs are often structured as pages, with each page presenting a specific field through pictures, audio, text or symbols. The student uses the structure that exists in these programs, selecting specific fields to activate one or more functions, such as reading a message, activating an MP3 player, writing letters, words or phrases, sending email or SMS, or activating spoken text or a calendar.

Rolltalk, *Tobii Communicator* and *Sarepta* can be used to make applications for communication, concept training, home assignments and educational materials. These tools come with examples, templates and prefabricated subprograms. The programs can be operated in different ways, with switches, joysticks, foot or head control, or adapted keyboards. These tools can be used for communicating through digitized or synthetic speech. The programs provide the student with bimodal choices and the student confirms or rejects messages, statements or options. The student may for example use a keyboard or a switch or produce vocalization to confirm an option, for example:

Level 1. *I want to drink / eat / go to the toilet*

Level 2. *cold / hot / warm*

Level 3 *water / juice / milk*

Level 4. *from a glass / using a straw*

A digitized or synthetic voice will confirm the student's choices. In the example above, the use of the hierarchical structure inherent in the program enabled the student to express «I want to drink – cold – milk – using a straw» through a sequence of choices. Using these tools without vision may be difficult, they make demands on the working memory capacity, because expressing the wish is based on a series of consecutive actions. There is thus a need to develop communication tools that are more tailored to the abilities and challenges of persons with JNCL.

Sarepta

In 1994, Statped (<http://www.statped.no>) in Norway initiated a collaborative project with Sprida Communication Center (<https://www.regionorebrolan.se/sprida#>) and Ekeskolan (<https://www.spsm.se/om-oss/english/>) in Örebro in Sweden to develop a computer program for students with JNCL. The first program was ground-breaking in many ways. It was among the first to use digital audio, images and synthetic speech. The program was first used by students with JNCL in Norway and Sweden and has since then gone through a long process of development in close cooperation with students, teachers and parents in several countries. The program was named Sarepta (Ragnarsson & Björk, 2009).

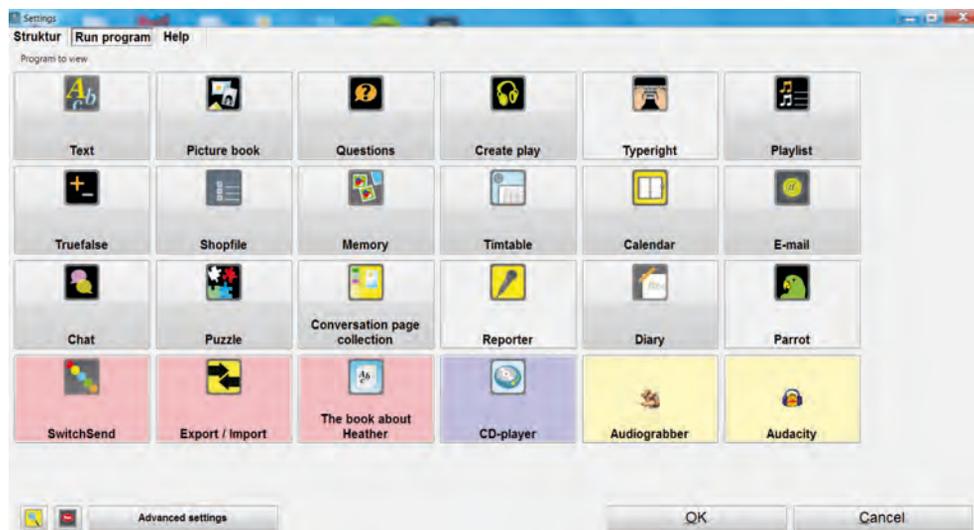


Figure 19.2 Modules in Sarepta

Sarepta is available in Danish, Dutch, English, Finnish, German, Norwegian, Sami and Swedish.

Sarepta is a shell program that makes it possible to tailor solutions for students who lack educational materials in school (Tengs & Torgersen, 2012). Teachers and residential staff fill the program with relevant educational material, which the students can subsequently make use of. The program is a multimedia tool with possibilities to mix audio files, video files and text files. Sarepta has been under continuous development and considerable efforts has been put into motivation-generating activities with new technological solutions, such as integrated digital and synthetic speech.

Sarepta consists of several modules, which give access to digital memory banks, texts, a media player, shopping lists, emails, a diary, picture books, playlists, memory games, quizzes, timetable, the book about me, logbooks, and so forth (see Chapters 20 and 23). The modules provide interactive exercises with feedback on assignments. Direct feedback is important for students with JNCL. The program includes a multitude of tasks and different ways of working, both individually and in groups with peers. The navigation of the program is simple and does not require literacy skills. Students have access to the modules and folders through bimodal choices (yes or no).

All the modules in Sarepta can be controlled by a simplified system with up to five switches (see section below). These five switches can be used to control external applications such as *Point braille*, *ABC* or other games (see Chapter 21).

Digital books

Digital books can be created with several computer programs and apps such as *Book Creator*, *Picture book in Sarepta*, *Phonto*, *Pictello*, *Album App* and *Clicker*. Some are developed specifically for people with visual impairments but not all of them are suitable for individuals with JNCL. These programs allow students to present themselves, their activities, interests, surroundings and background. The students can build a memory bank and store self-produced files with text, sound, film or photo. The following story illustrates how digital books may be used to encourage learning, motivation and inclusion:

“John” had a vocabulary below average and difficulty reading long words. Together with the teacher, he recorded sounds and made a series of digital images from a visit to the zoo. After the trip, John wrote a text describing his experiences on the trip. The audio recordings were used as sound illustrations, the supporting text was recorded digitally together with other students and edited with help from the teacher.

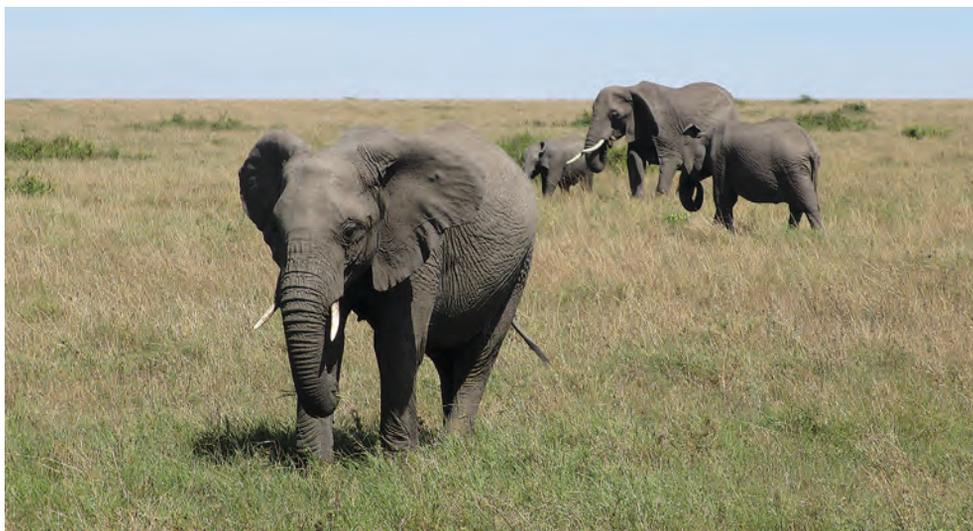


Figure 19.3 Picture book «The Elephant»

In the Picture Book, a student can write about a specific subject, such as this book about the elephant, which includes detailed information about elephants, physical traits, eating habits and geographical distribution, as well as stories and myths about the elephant as «the elephant never forgets». The student can later study the information and revise previous findings.

Digital books can also contain facts and information about school subjects and topics, such as «The elephant» with texts, images and sounds (Figure 19.3).

It is also possible to create digital books about the student's specific interests or hobbies. If the student is interested in auto racing, one can scan images and download sound tracks, for example from a Formula 1 race, and then add information about car producers, countries of origin, names of drivers, number of employees to expand the student's knowledge within his or her personal area of interest.

The Book About Me

The *Book About Me* (a module in Sarepta) is a written narrative of the person's life history (further described in Chapter 23). The *Book About Me* can be created as a digital multimedia platform with texts, audio recordings, photographs and films (like digital books above), for collecting and presenting information about the person. It can contain facts about the person, important events, daily activities, school attendance, projects, prizes and appointments, family, friends, neighbors, special interests, hobbies, and a map of all the places where the person has lived or visited.



Figure 19.4 The Diary module in Sarepta

The diary module in Sarepta is filled with information, events, activities, hobbies, adventures, experiences and topics selected from the person's personal experiences and interests. The structure of the program makes it easy for both students and staff to attach written materials, video clips, photographs, audio recordings and multimedia displays.

The Diary

The digital diary (also a module in Sarepta) is a program based on the standard calendar as shown in Figure 19.4 (see also The Yearly Wheel in Chapter 23). It can be operated independently using a braille keyboard with individualized switches (Tash, joystick, concept keyboard, etc.) or with assistance (interdependence, see Chapter 16).

The diary provides a range of possibilities, such as composing daily narratives or using preassigned categories and topics. It includes user-friendly editing functions like delete, save, undo and print located on a toolbar. It is possible to create a workbook, a logbook or a hobby book, where the student can store favorite movies, eBooks and music. A search function makes it easy to find special topics, dates or events. The diary can be used as a basis for conversations and social interactions with parents, relatives, friends or staff. The book may become an important part of the *Book about (the name of the person)* (see chapter 23).

The use of the digital diary may support students' development and maintenance of literacy skills, communication and ability to express wishes, thoughts, needs, feelings and experiences. Another user-friendly diary app is *Day One* (<https://dayone.me/>), which is simple to use and its content can be synchronized with *iCloud* or *Dropbox*.

Digital conversation sets

The idea behind the programs like *Talking Mats* (<https://www.talkingmats.com/>), *Notes* (<https://www.get-notes.com/>) and *Proloquo2go* (<https://www.assistiveware.com/>) is to support individuals with expressive communication problems. These programs can help individuals with JNCL express opinions related to selected themes. In *Talk* and similar tools, all statements can be answered with yes or no. Phrases that can help meet the identified needs of the person are recorded by teachers or others who know the person well. The programs can be operated by switches where the person listens and selects a phrase or option. Supporters can assist the person with JNCL by scrolling through the various options (see partner-assisted auditory scanning, Chapter 13).

Digital Logbook

A digital logbook may be used for communication between staff members and staff and parents (see Chapter 23). A log book can be used to record events of special importance, for example to describe happenings in the classroom or to communicate problems associated with epilepsy.

Social Media

Social media play an increasingly important role for youths in the modern society, and are important for social contact and communication (Mehraj, Bhat, & Mehraj, 2014). Some social media can be adapted to meet the abilities and challenges of individuals with JNCL. Comments from parents and staff in the present study (Appendix A) indicate that some individuals were ardent users of Internet resources like Facebook, Instagram, YouTube, Chatting and Snapchat with support from parents and staff. Social media make it possible for people with JNCL to keep in touch with acquaintances, friends and family and to share activities with their peers.

Some persons with JNCL have created their own profile on Facebook, with access for their friends and family. These persons keep in touch with friends with or without the use of writing. Written materials on Facebook can be read with a braille screen reader or synthetic speech. The person can get help to write comments or updates of any kind.

Instagram consists mainly of photographs and videos and may therefore be less relevant for persons with JNCL, although it may be used with interdependence where somebody describes the visual content (see Chapter 16).

Smalltalk and chatting play an important role in youth interactions, and since the 1990s, some persons with JNCL have used Chatting, a specially designed program which also includes synthetic speech output (<https://www.>

regionorebrolan.se/sprida). The chat groups are relatively shielded and accessible only to members of the group. Chatting has functions such as automatic saving and printing in print or braille. It may be useful in education, for example with quizzes and discussions around special themes, as the following story illustrates:

A girl with JNCL organized fund-raising activities when she was a child. Two times a year she organized a bazaar to collect money for a particular child in Africa, and made her bazaar known through the social media. This activity occupied the girl throughout the year and she involved family, friends and people in the neighborhood. She was the head of the organizing team, collected items, asked for support from the local bank, wrote letters requesting sponsorship, managed the bazaar with support from others and finally sent the money to the child in Africa. The girl's motto was «There are a lot of children who have a worse situation than me!»

Skype, Viber and S-Chat are other examples of social media adapted for persons with visual impairment. Being engaged in social media may support literacy skills and contribute to social well-being.

Taking pictures

Photography is not usually a special interest of children who are blind, but there are exceptions. The story about this young man with JNCL illustrates that a positive and open mind can lead to astonishing results by using creativity:

“Jack” was very interested in taking photos whilst he still had some vision left and kept this interest after he became blind. He took photos together with his family and it was a very exciting activity for the boy. After a few years, the parents started to carefully study these photos and discovered that many of them were good and special. Sometimes Jack would miss the motive and the photos would have a new content and other artistic dimensions. He challenged himself to learn photography using other senses – being more attentive to sounds and tactile qualities. For example, by listening to someone talking he could more or less assess the distance and height he needed to aim for. The family had great fun when watching and discussing these photos and lots of humor was triggered by the pictures.

The parents presented the photos to the well-known Norwegian photographer, Morten Krogvold, who was so impressed with Jack's artistic pictures that he set up an exhibition with Jack's photos at the Nordic Light-festival in the Norwegian city Kristiansund. The exhibition was a huge success and received positive reviews in the media. It was a great experience for Jack and his family. His story is now spread on social media.

Jack's story shows that early interests – in this case photography – can be maintained with support even if a decline – here blindness – gives the activity a new and unconventional dimension.

Email and SMS

Most people send and receive emails and SMSs, and these tools can also be used by persons with JNCL for keeping contact with peers, family and society. The use of email and SMS may contribute to maintaining friendships when physical meetings with friends are limited. The following example shows that email correspondence can strengthen participation in society:

A few years ago, a young man with JNCL corresponded with family and friends across the country via email. He had an assignment to make his own electronic newspaper about football which was distributed via email. He surfed regularly on the websites of Manchester United (football club in UK) and Rosenborg (football club in Norway) to catch the latest news of these clubs. He also made interviews with his favorite players in these clubs, which were published in his newspaper. The paper became very popular.

Speech recognition may be of great help when young people with JNCL use email and SMS when their speech is intelligible. However, *interdependence* or the *Team Model* will be needed when speech becomes unintelligible (see Chapters 16 and 23).

Many assistive technology programs have integrated email systems with automated storage features. The person with JNCL can record their own voice or speak directly in the email and send the recordings as digital files. Email systems can be operated by switches or keyboard. Comments from parents and staff in the present project indicate that active use of email was motivating and contributed to the maintenance of writing and reading.

Multimedia equipment and text editors

With multimedia editor programs, persons with JNCL can combine files with audio, text, images and video. It is possible to create individual material which enables students to adapt their learning efforts to their own learning progression and regression. The possibility of using sound, switch control, screen magnifier programs and braille display makes digital solutions available to persons with JNCL.

The Braille Cell

The Braille Cell (<https://www.regionorebrolan.se/sprida>) is a computer program with audio support designed for teaching braille at an early stage. It may support the student's understanding of the braille cell structure by using a regular keyboard or a braille keyboard (see Chapter 14). The program produces letter sounds or spoken words as the braille cells are produced (Figure 19.5). The program includes, in addition, an interactive part named *The Adventure*, with several premade adventures. The student can work with these adventures to facilitate literacy skills. They can also create their own adventures in *The Adventure*. The Braille Cell can be used in parallel with applications such as *Braille Hunt* (<http://www.textax.se/>) and materials for *Flexiboard* (<https://www.abilia.com>).

Alternative keyboards

Alternative keyboards, such as *Flexiboard* (see Figure 19.6), can be adapted to a person's special needs and may enable students with JNCL to use the computer. The board has several layers of tactile material which the student can identify through touch. It is possible to add recorded speech or other sounds. *Flexiboard* has many other applications for leisure activities and schoolwork. For example, the student can be given an overlay with the map of Finland. By touching different areas of the overlay, the student will be given information about different places, for example *Helsinki is the capital of Finland and has 620.000 inhabitants*. Map use can be expanded to include topics for conversation, including telling others where one has traveled, or where friends and family live, and then inviting others to expand on those ideas. *Flexiboard* can support learning in all school subjects, for instance in biology or music. It is possible to create a multimedia presentation by using *Flexiboard*, for example include speech, music and videos in one presentation.

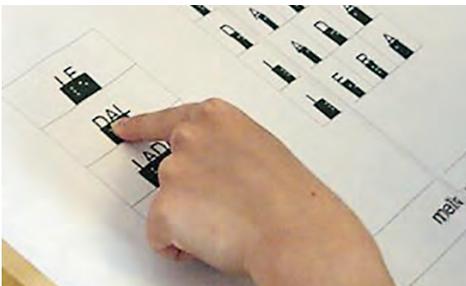


Figure 19.5 The Braille Cell

The user gets audio feedback as the braille cell is built.



Figure 19.6 The Flexiboard

The flexiboard offers more possibilities than the traditional keyboard.

Aids to support writing

Some aids support the process of composing one's ideas in print. These are typically apps or programs, not hardware (see Batorowicz, Missiuna, & Pollock, 2012). Many such programs, including *Ghotit* (<https://www.ghotit.com/>) and *Lindys* (<https://lingit.no/produkter/>), are designed to help students with reading and writing difficulties (MacArthur, 2009). They are mainly used by students with dyslexia, and some students with JNCL can benefit from using these programs, especially in primary school. For example, if a student can write the first letters, the program can predict a number of words that begin with these letters. The student can then select the right option. This function is common in text messaging. Two types of support are provided: writing might be quicker with fewer keystrokes, and it can compensate for difficulties in spelling and memory. The combination of spelling support, a built-in dictionary, word prediction, multimodal input, screen

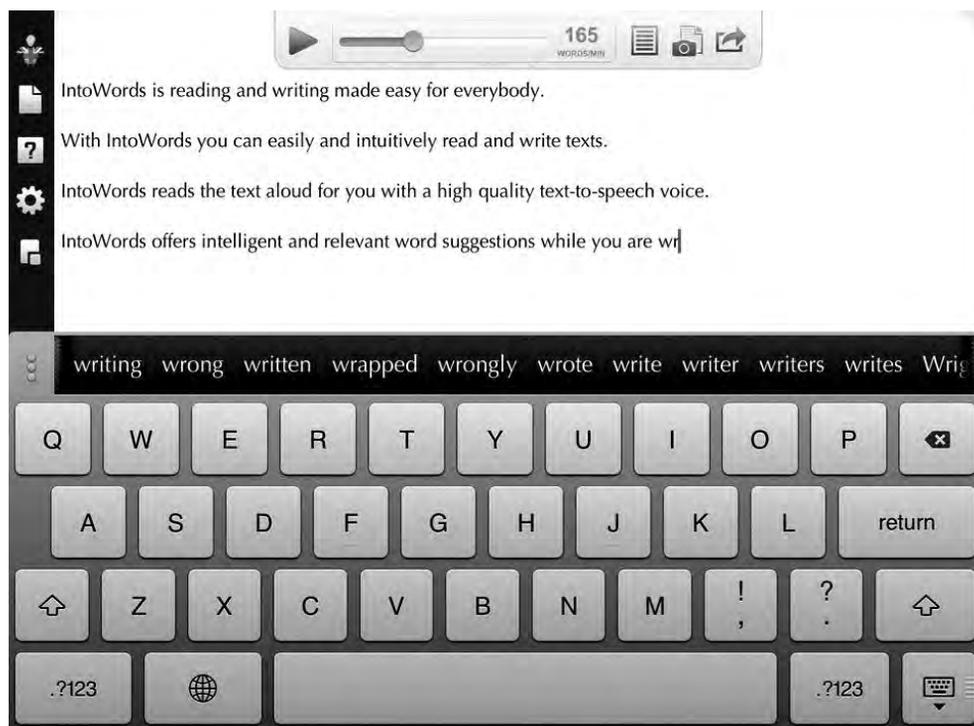


Figure 19.7 IntoWords is a reading and writing support tool

IntoWords is a digital tool for reading texts aloud and compensating for reading and writing difficulties. It uses word prediction, helping the student by suggesting the next word.

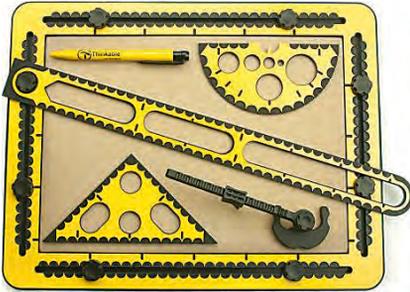


Figure 19.8 TactiPad with drawing tool

TactiPad is a versatile drawing board, which enables a visually impaired person to make a tactile drawings by firmly pressing down on the foil with a pen; the lines will raise instantly, forming a tactile image.

reader and synthetic speech output makes *Textpilot* (<https://lingit.no>) useful for working with texts.

The app *IntoWords* (<https://www.mv-nordic.com/en/products/intowords>) also provide writing and reading support (Figure 19.7). *Clicker Grids* (<https://www.cricksoft.com/>) may help students learn the relationship between written and spoken words. The synthetic speech feedback allows students to review and take control of their own writing (Genlott & Grönlund, 2013).

The app *WordWav* converts text into an audio file which can be read aloud on different audio players. This makes *WordWav* a useful tool for some students with JNCL.

The TactiPad

The TactiPad is a versatile drawing board, which enables persons with visual impairment to produce tactile drawings (Figure 19.8). These drawings are made on a thin sheet of plastic foil on top of a layer of rubber. By firmly pressing down on the foil, the lines will rise instantly, forming a tactile image (<https://irie-at.com/product/tactipad-drawing-tablet/>). The technology is similar to swell paper (see below). The drawings might be used to support the student's own written texts, or substitute for them. One can also communicate ideas through drawings.

Digital recorders

Digital tools, such as digital cameras and voice recorders, are useful in education. During visits to museums, Christmas workshops or outdoor fieldwork, teachers and students can work together to record the activities, for example by taking photographs or making video and audio recordings. These creative products may allow the student to relive the activity, refresh memory and create a basis for conversations, discussions and school assignments.

Students can use an ordinary audio recorder or apps to record television or radio programs, speech or other sounds in their surroundings. Students may for example record their thoughts and opinions about events during the day, which can be included in their diary.

The use of a digital recorder within and outside the classroom may facilitate learning and motivation. The *Reporter* module in *Sarepta* (see above) functions as a sound recorder that can be used when interviewing people. This module is a simple digital recording app which includes speech synthesis. Recording can be controlled by two switches (on and off) or with F4 and F5 keys on a standard keyboard. The files are automatically saved in the audio bank in MP3 or WAV format, and recording time is limited only by the size of the hard disk of the computer. Recorded audio files can be deleted or edited by numerous media programs, such as Microsoft's *Sound Editor* or *WavePad*.

The student's library

With the help of programs such as Audacity (<https://www.audacityteam.org>), students with JNCL can build up a library of self-produced texts, audio recordings, audio books, radio programs, and so forth.

Music

Music is a source of pleasure and of special importance for individuals with JNCL according to the present study (see Chapters 17 and 18). Active participation in singing, playing an instrument and listening to music together with others, provides social and cognitive stimulation and facilitates inclusion. Comments from parents and staff in the present study indicated that music is a life-long interest for many persons with JNCL (see Chapter 17).

There are a number of computer programmes and music technology products which parents or staff can utilise to engage children and young people with JNCL in music activities.

Programmes on the iPad such as *Echostring*, *Drumbeats* and *Sound Prism* have been shown to be successful for individuals with some range of movement in their hands or arms, and for whom conventional instruments are too difficult to play. One parent commented on her son's enjoyment of *Drumbeats*.

It has all the instruments that he likes. He loves the ocean drum and when you tip the iPad, it makes the same sound. This is brilliant because he cannot really hold the real ocean drum as well anymore, but the iPad he can still grip and tilt.

These interactive programmes allow children and young people to make sounds easily and help develop an awareness of how they can use small movements in their hands to make interactive sounds. However, they are too limited for children and young people who have the cognitive ability to engage in more complex music activities, such as song-writing or music improvisation. For this group, programmes such as *Garageband* and *Launchpad* may be used in school and at home, to encourage children to execute decision making, planning, and enjoyment in making their own music.

The *BEAMZ* unit, an interactive laser-beam console, has been trialled with individuals who have a good awareness of the cause and effect of movement, but who have limited gross motor skill in the upper or lower body. Children and young people with a higher cognitive ability have navigated their way around the console using their fingertips to determine where the lasers are situated and how sounds can be manipulated by movement. They have also recorded pieces of music and listened to these being played back to them.

Makey Makey is a system which can turn everyday objects into touchpads and combine them with the internet to play games, music or create switches.

Audio books

Audio books represent an important compensatory medium for people with severe visual impairment or reading and writing difficulties (Saine, Lerkkanen, Ahonen, Tolvanen, & Lyytinen, 2011). Listening to audio books supports social participation by supplying topics for conversation. It is also a meaningful leisure activity, as reported by parents in the present study (Appendix A). Many books and radio programs are available as audio books. Audio books give people with JNCL access to a large selection of fiction, nonfiction, academic books and newspapers. Audio books are also available in many libraries. In addition, there are many applications for purchasing and downloading audio books on internet, such as *Storytel* (<https://www.storytel.no/>), *E-book* (<https://www.ebooks.com/>), and *Audio Book* (<https://www.audible.co.uk/>). The national associations for persons with visual impairment produce audio books in order to provide equal access to literature and information. These services are free and nationwide in many countries.

Electronic tools for daily living

Time management is important in modern societies, but understanding time and reading *digital clocks* can be difficult for many persons with JNCL. There are watches with synthetic speech that can be adapted to individual needs. It is possible



Figure 19.9 – A QR code

QR codes can store any kind of information. They can be used to identify personal objects like books or CDs, or to download digital versions of audio books on Internet.

to add personal messages to some of these watches. *Cell phones* can be used in many different ways and may be adapted to the student's abilities and needs. They may provide support for time, planning, structure and memory. The calendar on the cell phone can show planned activities and alert the person when an activity is starting or ending. *SmartVision* (<http://www.sightandsound.co.uk/>) is a smart phone that is designed specifically for people with visual impairment. This type of phone is suitable for children and adults with JNCL. *SmartVision* combines a physical keypad, a full voice interface, and a touchscreen. It can be organized according to personal preferences and is user friendly, with a large display, large icons, four font sizes, tactile buttons for numeric keys and navigating, voice commands and speech-to-text facilities. It can receive and send text and audio messages.

A *digital schedule* provides the same features as a normal calendar but may have a range of further possibilities. The schedule can be supplemented with personal information and text, images and audio. For instance, a gym program can be specified in the digital schedule and provide information about the plan for the next day. A digital schedule (and other devices) can be used for making shopping list, check list and so on.

A QR code (quick response code) is a matrix barcode, which can contain comprehensive information about an object, for example in supermarkets (Figure 19.9). The QR codes are generated online and can be read by QR scanners on the cell phone, and apps that utilize QR technology are readily available for desktop and mobile applications. QR codes can be useful in education as they can store all kinds of information, such as week plans, shopping lists, mathematics and biology. QR codes can be used to identify personal objects like books or CDs, or to download digital versions of audio books from Internet.

Pocket-sized GPS navigation devices with voice recognition and synthetic voice output, like *Captain mobility*, *Breeze* and *Blind Square* are practical orientation and navigation aids for persons with visual impairment (Zegarra & Farcy, 2013). These and similar GPS-based tools are able to provide information about current location, the streets at the next junction, places of special interest, shops, restaurants, pharmacies, and more. Some of these aids have information about buses, trams, trains, subways and other forms of public transportation. For

instance, when the user clicks the *Where am I* button, the device gives information about the street name of the current location and the travel direction. It is possible to add personal points of interest into some of the devices. It is not known if any of these technologies are used by persons with JNCL, but they might be perceived as fun and exciting by some individuals with JNCL.

Maps and atlases can be useful when talking about places, or to find the name of a town or country. There are various map applications to choose from, including *Google Maps* and *Google Earth*. It is possible to use speech input to navigate and synthetic speech output with these maps.

The *Mobile Reader* (<https://acrobat.adobe.com/>), *TapTapSee* (<https://taptapseeapp.com/>) and similar apps installed in cell phones have scanner, speech and reading functions. The scanner function makes it possible to take pictures of documents like bills, menus and banknotes. The image is transformed to a spoken description which can be saved on the phone. It is not known if such devices are presently used actively by individuals with JNCL.

The PenFriend (<https://www.rnib.org.uk/>) is an audio labelling system for individuals with visual impairment. The system consists of a digital pen and labels which contain readable barcodes. The pen has a scanning and memory function, a microphone and speaker system. Each label can be combined with an audio recording. PenFriend can be used for cooking instructions, dietary information or the date when something was placed in the freezer. It can also be used to keep track of personal items like CDs, DVDs and books.

Assistive technologies such as the *Tactile Image Maker* (<http://www.perkinselearning.org/>) make touch-based drawings on special swell paper. The student can draw, print or photocopy pictures onto a swell paper and pass it through a swell machine. The heat from the swell machine makes the illustration readable with touch (see <http://piaf-tactile.com/producing-tactile-graphics/>).

Similar assistive technologies allow persons with JNCL to make drawings and design or read simple graphics, maps and similar. This type of technologies may be useful for persons with JNCL, both in education and leisure activities, and may promote inclusion when persons with JNCL are taking part in classroom activities with peers.

Technologies like *Loc-Dots* (<https://www.independentliving.com/>) consist of raised dots to be used for tactile marking. The dots are simple to use and can be placed where needed, for instance for marking of switches, the computer keyboard, books and tape recorders. Such technologies may simplify daily living for individuals with JNCL.

There are several computer programs and apps available for doing crossword puzzles, memory games, board games, chess, lotto games or card games. Some of these games may be used to enhance learning and pleasure for children with

JNCL. Examples are *Scrabble Braille*, *Monopoly Braille*, *Arcade* and *Digital Memory Lotto* (<https://shop.rnib.org.uk/>) (see Chapter 21).

Persons with JNCL who have severe motor impairment are often dependent on switches to control computers, communication devices, cell phone and other devices. It is important that students with JNCL are introduced to such switches at an age when their learning capacity is at its best, according to the principles of enhanced and proactive learning (see Chapters 11 and 12). Moreover, care should be taken in switch selection, so that the properties of the switches match the sensory and cognitive capabilities and preferences of the switch user. Switch technology now accommodates a very diverse range of users (Gibson et al., 2013)

Careful consideration must be given to the quality and quantity of the feedback a switch affords. For example, many switches make a clicking sound to notify the user that a "click" has been activated. Switches have a range of tactile properties to cater to tactile preferences, in the case of people who have atypical tactile sensitivity, or tactile needs, as in the case of someone who requires considerable tactile feedback. Vibrating switches, for example, provide constant feedback during the entire time the switch is activated. Over the course of the period of decline that accompanies JNCL, there will be a need for re-evaluating the type of switches used and the provision of individual support.

Recommended computer equipment and other aids

Recommendations for assistive technologies for persons with JNCL should always be based on an assessment of needs, functions and possibilities (see Chapter 10). Assessments must be made by professionals who have sufficient competence related to JNCL and assistive technology. Assistive technologies are often expensive, and comments from parents and staff in the present study indicate that many of the devices that were provided never had a positive impact on the learning and well-being of the person. The reasons were often a lack of local knowledge about the devices and an over- or underestimation of the functional use of the device.

A core device for a person with JNCL is a laptop computer with large storage capacity. Files with audio, images and video require extensive memory capacity. A good sound card is mandatory, and appropriate switches or other input devices for operating the electronic equipment. Braille keyboard and display are often needed and a Flexiboard may serve as an adaptable keyboard and mouse. High-quality audio equipment plays an essential role for persons with JNCL, including a high-quality microphone. The device should have high quality speech synthesis, for several languages if the person is living in a multilingual environment or learning

a foreign language in school. A portable multi-player with optical character recognition (OCR) and text-to-speech for scanning books and documents is also a basic equipment for individuals with JNCL. The devices should have a numeric keypad and a dictionary with possibilities for doing calculation and making memory notes. The device should be able to play audio books and music files from internal or external memory devices and should be supplied with an earphone that turns off the external speaker when connected, to avoid disturbing others.

A digital camera may be used for educational and social purposes. A braille printer, located where it does not disturb others, is essential for braille users. A color detector is easy to use, and has a wide range of useful applications, from checking the color of clothes to the ripeness of fruit.

References

- Anusuya, M. A., & Katti, S. K. (2009). Speech recognition by machine, a review. *International Journal of Computer Science and Information Security*, *6*, 181–205.
- Bandura, A. (1997). *Self-efficacy: The exercise of control*. New York, NY: W. H. Freeman.
- Bandura, A. (2006). Toward a psychology of human agency. *Perspectives on Psychological Science*, *1*, 164–180.
- Batorowicz, B., Missiuna, C., & Pollock, N. (2012). Technology to support written productivity in children with learning disabilities: A critical review. *Canadian Journal of Occupational Therapy*, *79*, 211–224.
- Buli-Holmberg, J., & Ekeberg, T. R. (2008). *Likeverdige og tilpasset oppl ring i en skole for alle* (Equal and adapted education in a school for all). Oslo: Universitetsforlaget.
- Carey, A. C., Friedman, M. G., & Bryen, D. N. (2005). Use of electronic technologies by people with intellectual disabilities. *Mental Retardation*, *43*, 322–333.
- Cook, A. M., & Polgar, J. M. (2008). *Cook and Hussey's assistive technologies: Principles and practice, Third edition*. Philadelphia, PA: Elsevier.
- Craft, A. (2012). Childhood in a digital age: Creative challenges for educational futures. *London Review of Education*, *10*, 173–190.
- Cull, W. L. (2000). Untangling the benefits of multiple study opportunities and repeated testing for cued recall. *Applied Cognitive Psychology*, *14*, 215–235.
- Edwards, L. C. (2007). Children with cochlear implants and complex needs: a review of outcome research and psychological practice. *Journal of Deaf Studies and Deaf Education*, *2*, 258–268.
- Freitas, D., & Kouroupetroglou, G. (2008). Speech technologies for blind and low vision persons. *Technology and Disability*, *20*, 135–156.
- Genlott, A., & Gr nlund,  . (2013). Improving literacy skills through learning reading. *Computers and Education*, *67*, 98–104.
- Gibson, B. E., King, G., Kushki, A., Mistry, B., Thompson, L., Teachman, G., Batorowicz, B., & McMain-Klein, M. (2013). A multi-method approach to studying activity setting participation: Integrating standardized questionnaires, qualitative methods and physiological measures. *Disability and Rehabilitation*, *36*, 1652–1660.

- Henderson, S., Skelton, H., & Rosenbaum, P. (2008). Assistive devices for children with functional impairments: Impact on child and caregiver function. *Developmental Medicine and Child Neurology*, 50, 89–98.
- Jana, R. (2009). How tech for the disabled is going mainstream. *Business Week*, 4149, 58–60.
- MacArthur, C. A. (2009). Reflections on research on writing and technology for struggling writers. *Learning Disabilities Research and Practice*, 24, 93–103.
- Mehraj, H. K., Bhat A. N., & Mehraj, H. R. (2014). Impacts of media on society: A sociological perspective. *International Journal of Humanities and Social Science*, 3, 56–64.
- Petty, R. E., & Frieden, L. (2012). *Technology access in the workplace and higher education for persons with visual impairments: An examination of barriers and discussion of solutions*. Houston, TX: Independent Living Research Utilization at TIRR.
- Ragnarsson, T., & Björk, J. (2009). Sarepta – a powerful multimedia program. Presented at the 7th European Conference of ICEVI, Dublin, Ireland, January, 2009.
- Saine, N. L., Lerkkanen, M. K., Ahonen, T., Tolvanen, A., & Lyytinen, H. (2011). Computer-assisted remedial reading intervention for school beginners at risk for reading disability. *Child Development*, 82, 1013–1028.
- Scherer, M. J., & Glueckauf, R. (2005). Assessing the benefits of assistive technologies for activities and participation. *Rehabilitation Psychology*, 50, 132–141.
- Shadiev, R., Hwang, W.-Y., Chen, N.-S., & Huang, Y.-M. (2014). Review of speech-to-text recognition technology for enhancing learning. *Educational Technology and Society*, 17, 65–84.
- Shanmugapriya, P., Gayathri, R., & Deepa, V. (2016). Android application to student education monitoring device range and real-time performance tracking. *International Journal of Information Technology and Mechanical Engineering*, 2, 1–7.
- Silver-Pacuilla, H., & Fleischman, S. (2006). Technology to help struggling students. *Educational Leadership*, 63, 84–85.
- Sølvberg, A. M. (2003). IKT i skolen: en studie av relasjoner mellom bruk av IKT på 8. klassetrinn og noen motivasjonelle faktorer (ICT in school: A study of relations between use of ICT in 8th grade and some motivational factors). Trondheim: Fakultet for samfunnsvitenskap og teknologiledelse.
- Tengs, A. M., & Torgerstuen, K. (2012). Å gjøre det uforståelige forståelig. IKT som pedagogisk hjelpemiddel i undervisningen med elever med døvblindhet (Making the incomprehensible comprehensible. ICT as a pedagogical aid in the education of student who are deafblind) *Spesialpedagogikk*, 77, 4–11.
- Zegarra F. J., & Farcy, R. (2013). GPS and IMU (inertial measurement unit) as a navigation system for the visually impaired in the cities. *Journal of Assistive Technology*, 7, 47–56.

20

Quiz Games for Building and Maintaining Knowledge

Mohammed Beghdadi and Bengt Elmerskog

Quizzes are multiple-choice games or mind games that are becoming more common in education in many countries. Quizzes are usually scored in points and many quizzes are designed to determine a winner, a single person or group. Many quiz platforms are available online, such as Kahoot and Triviaplaza, and these can provide ideas and resources for teachers and students, when there is a need for quizzes with an educational focus. Quizzes lend themselves to many formats allowing teachers to use them as a class activity or to structure materials for students to use in self-study to repeat reviewed material. Teachers sometimes encourage students to design quizzes themselves as a review strategy with a competitive twist.

Quiz in education

In school, the quiz has a long tradition. Asking everybody in the class specific questions is a standard teacher activity, as well as using tests with multiple choice answers or questions that require a short answer to evaluate the knowledge of the students (McDermott, Agarwal, D'Antonio, Roediger, & McDaniel, 2014). Most quizzes in education are developed locally to support predetermined teaching goals. Many quizzes are programs that can be performed on a computer, tablet or smartphone, but few such quiz programs are adapted to meet the special needs of students with visual impairment and cognitive disorders.

Using quizzes can function as an alternative or complementary method to achieve and support different educational purposes or teaching goals (Brusilovsky, Sosnovsky, & Shcherbinina, 2004). A quiz can provide multimodal input and repetition that may contribute to improved learning. They can be used as a variation from the traditional forms of teaching and learning. Many students perceive quizzes as fun, and the questions may stimulate and promote new interests. The

teacher can use quizzes to assess the students' knowledge of a particular subject or theme, or the students' ongoing growth in knowledge, abilities and skills. Students may use quizzes to test themselves and get feedback on their own learning process. For instance, a quiz may be used to check the comprehension level of a type of mathematical exercise in a class. Quizzes are often presumed to be less formal, more short and concise, and easier to organize than other forms of written tests and examinations.

Quizzes can be used to *refresh* the content of one or more lectures. The students attend a lecture and take a short quiz on the lecture content afterwards. Such use of quizzes can also function as a student response system that can be used to guide the teaching (Melero, Hernández-Leo, & Blat, 2014). It can measure what and how students are thinking and address the needs immediately in class. Quizzes are also used to check students' prior knowledge, probe their current understanding, and uncover student misconceptions. They can provide feedback to the teachers about their students' understanding and to the students about their own comprehension.

Some schools schedule a daily or weekly quiz for the purpose of having the students review recent lessons before attending the next class. Further, quizzes are used to check if the students have done their homework according to the plan. Using a "pop quiz" means that the teacher gives it without warning: students are not given time to prepare for the task, and so they are taken by surprise.

Learning facts and making personal reflections

Quizzes are most appropriate for learning facts, for instance within geography and history, or learning by heart, for instance the multiplication table. Quizzes are less appropriate for learning things that require certain insights, appreciations, judgments or reflections. However, further elaboration around questions presented in a quiz game may emphasize further insights. It is often motivating for the student to compare answers with others while waiting for the results. Such peer collaboration may contribute to further insight and to the students becoming aware of different aspects of the presented tasks. This may stimulate curiosity and learning because it may lead to discussions involving personal experiences and emotional engagement. The teacher can in addition use the interest caused by the quiz to present further elaboration and explanation. The quiz may be used strategically as a door opener or a starting point for deeper understanding and further capacity and competence building in predefined domains.

Refreshment and consolidation of learning

Retrieval practice is a common teaching strategy where the teacher introduces actions and activities to consolidate the student's knowledge (Chang, 2015). The student is practicing exercises to activate what is already learned as opposed to acquiring new knowledge. The act of retrieval, or calling information to mind, will refresh the memory and make forgetting less likely to occur (Morey & Cowan, 2018). Retrieval practice is a powerful educational strategy to consolidate knowledge and improve the effect of learning (Uner & Roediger, 2018).

Some authors recommend use of quizzes as vehicles for providing repeated retrieval practice (Karpicke, Butler, & Roediger, 2009; Karpicke & Bauernschmidt, 2011). Learning is typically identified with acquisition, encoding or construction of new knowledge, while retrieval is considered a means for accessing knowledge, rather than a process that contributes to new learning. The process of consolidating new knowledge in a strategic way is not always emphasized in education. However, some authors suggest that retrieval is a key process for both understanding what has been learned and for promoting learning, yet retrieval is not granted the central role it deserves. Research, particularly that of Karpicke and colleagues, shows that closed-book repeated quizzes have better impact on long-lasting learning than open-book questions. Existing educational practices can easily be converted into retrieval-based learning activities according to the authors, for instance by answering questions by taking repeated quizzes. Classroom quizzing is one effective method among other methods for effective implementation of retrieval practices (Agarwal, Karpicke, Kang, Roediger, & McDermott, 2008). The importance of retrieval practices holds true even in preschool populations. During the preschool years, children are learning vocabulary, but some children have difficulty relative to their peers and require support. Activities that provide retrieval practices are thus valuable because even preschoolers learn vocabulary better when they are given such opportunities (Fritz, Morris, Nolan, & Singleton, 2007). This research might provide insight into ways that can promote enhanced retention of knowledge in children with JNCL in the face of decline.

Quizzes and teaching

Quizzes can be used by teachers to build up a positive atmosphere in the classroom, for example to reduce stress. Quizzes are often perceived by students as fun and relaxing if they are used in a balanced way; they can create feelings of pleasure and joy. Quizzes are rather informal compared to other forms of evaluation. They may have a competitive element that creates excitement among most children and

young people. The provision of multiple-choice answers will in addition give the student a fair chance to appraise appropriate answers by ticking the most probable alternative. Multiple-choice answers have the advantage of reducing some of the memory retrieval load: the correct answer is available, and it is up to the child to appraise the options, not generate them.

A quiz with multiple-choice options usually requires less time to administer than a test with written responses. The teacher does not need to interpret answers because students are graded purely on their selections. Moreover, clarity of presentation is not an issue when using an online or electronic quiz and poor handwriting will not influence the results, although the child or young person may accidentally mark the wrong box. An additional advantage is that the evaluation of a quiz makes it easy for students to acknowledge their own knowledge or lack of knowledge in a rather distinct way. Quiz tests can be strong predictors of overall student performance compared with other types of evaluations, but the complexity of the material and other factors may influence the learning effects of quiz tests (Adesope, Trevisan, & Sundararajan, 2017; van Gog & Sweller, 2015).

Quizzes for people with dementia

Quiz games are often used beyond education, in connection with festivity, celebration or other types of social meetings to create a good atmosphere. A quiz can easily be adapted to facilitate and adjust the content in accordance with individual needs and abilities, for instance for individuals with learning difficulties. Quizzes are used for elderly people with the diagnoses of dementia (Elder Care, 2016). Quizzes and similar games, like crossword and puzzles, are common activities among elderly people with or without dementia, since such games provide pleasant social settings and cognitive stimulation at the same time.

According to cognitive stimulation theory, quizzes are considered to be "brain games" that may prevent or at least delay the loss of knowledge and skills in connection with the progression of dementia (Dormann, 2016; Livingston et al., 2017). Quiz games may be a useful way for elderly people to exercise their mind. Questions about various subjects will get their mind working and stimulate parts of the brain that otherwise may not be so much used. Elderly people with dementia are found to do rather well in quiz games compared to many other cognitive games, partly because it is easier to recognize correct options than to recall information (see Chapter 5). Using quizzes on a regular basis may contribute to maintaining old memories of special importance for the person (i.e., reminiscing). Studies of reminiscence therapy suggest benefits to adults with dementia in the

form of mood and cognition improvement, and benefits to caregivers in the form of lower strain (Cotelli, Manenti, & Zanetti, 2012; Woods, Spector, Jones, Orrell, & Davies, 2005), especially when related to the life history and personal experiences of the individual (Subramaniam & Woods, 2012).

A strength of the use of quizzes for people with dementia is the immediate response to the answers that are selected (Balota, Duchek, Sergent-Marshall, & Roediger, 2006). Immediate responses require lesser demands on the capacity of the short-term memory. Keeping the mind stimulated is even more important in younger people with dementia disorders, and doing quizzes, puzzles or crosswords that invigorate the brain are highly recommended by experts. For instance, quizzes that incorporate numeracy can be useful for keeping those skills alive according to Alzheimer's Society in UK (2018).

Quiz use among children and young people with JNCL

In the present JNCL study (see Appendix A), quiz games were often mentioned by parents and staff as an activity of special interest for the participants with JNCL. Moreover, the interest for doing quizzes was described as a life-long interest for some individuals with JNCL. Both school teachers and staff in residential homes for young adults with JNCL said they were using quizzes as strategic tools to enhance learning and help the individual maintain memories and skills. Many parents reported using quizzes as a joyful leisure time activity to promote fun and well-being. The quotations below from parents and staff suggest that quizzes may have a special role in the life of children and young persons with JNCL:

He has always loved participating in quiz games. He makes his own music on YouTube, we make quizzes about music, for instance, who composed this song? These games are used for building up his self-confidence, he becomes very proud; playing quiz games is also a way to see if he is able to accomplish same tasks as previous years.

So like, for example, we were doing time and math, they focused on the calendar and we worked on the months of the year and we started off with how many months there are and – and we gave her the little rhyme that goes «30 days has November» and she brailled that out and then we had the little quiz with all the months and she had to say right she'd find, she'd take the month January for example, how many days does January have?

And she wouldn't know so we went through the rhyme again and that and she would work it out and put the right – find the right number and put it on the calendar, there was maybe a choice of two numbers.

We have all types of games, board games for the blind or self-made games. Sometimes we modify original games and adjust them to her mental progress. She prefers quizzes, she just loves it. She also loves the Tiptoi books. Her interest in games grew over time, games have been very helpful.

We use themes in life when preparing quizzes. You could talk about topics which concern and move them, more in the direction of life planning – with different levels. Some kids with JNCL could complete tasks by writing braille and they enjoyed it. This was just their competence level. With other kids we had to do a quiz game: «What's the capital of ... and which river passes through ...?» Then they answered and were very proud and happy. That was helpful. Or we utilized their knowledge of foreign languages. «Which language do you speak? What is 'good morning' in French?» and so on, that's all possible if it accords with the interest or knowledge the kids have.

Our child is very eager to learn. She learns a lot by doing quiz games (for children, Tipp Toi etc.) about general knowledge but especially about nature, environment and animals. She is very creative and again she emphasizes that she has an advantage over blind-born children.

We conclude every school week by doing a group quiz. It is a perfect way of concluding and recapitulating the week.

The most popular activities in the leisure time club are swimming, bowling and doing quizzes. Her performance level in doing quizzes is impressive.

The comments show that quizzes are used in the countries that participated in the present project to promote learning and maintain skills and knowledge, as well as to promote joy and excitement in children and young people with JNCL. However, both parents and staff expressed the view that quizzes could be used more, both for teaching and cognitive stimulation, and for stimulation and joy beyond school.

Quiz Tool for persons with JNCL

As part of the present JNCL and education project (Appendix A), a simple and automatically-controlled quiz program, the Quiz Tool, which has the necessary level of repetition of content, alternative answers and immediate responses was developed for children and young people with JNCL. This part of the project received extra funding from The Norwegian Directorate for Education and Training and the Norwegian NCL Family Association. The motivation behind the project was that a quiz program represents an alternative method for contributing to achieving new learning, to refreshment and conciliation of knowledge, and to maintaining cognitive skills and knowledge in children and young persons with JNCL. The Quiz Tool may be used by people with or without vision. The instructions can be read through vision (print), the tactile modality (braille), or hearing (speech synthesis).

The Quiz Tool can be used to create a variety of quiz categories, for example of school subjects such as geography or history, activities like playing or sports, or events such as birthdays and holidays (Figure 20.1). This can help the student with JNCL to get an overview of important events and activities. The Quiz Tool is flexible and enables teachers to teach subjects with questions tailored to each student's needs, desires and abilities. The tool can be used for most subjects



Figure 20.1 The Quiz Tool with different quiz categories

The desired quiz game will open by ticking the category. The selected category, for example «Adjective» can be read or heard with the speech output of the computer.

Table 20.1 Examples of questions used with the Quiz Tool

1. Standard quiz questions

Question: Which city organizes the annual Academy Awards, also known as the Oscars?

- A: New York
- B: Los Angeles
- C: Chicago

Question: When was Michael Jackson born?

- A: 29. august 1958
- B: 29. august 1956
- C: 29. august 1960

2. Incomplete sentence

Question: Norway's annual international Fish Convention is organized in:

- A: Oslo
- B: Bergen
- C: Trondheim

3. Grammatical error

Question: Which sentence is correct?

- A: I saw a football match yesterday
- B: I saw football match yesterday
- C: I see a football match yesterday

4. Quiz questions requiring different focuses

Question: The town Tromsø in Norway

- A: is located in Nordland county
- B: has more than 250 000 residents
- C: is described as Paris of the Scandinavia

5. The Quiz Tool can be used with daily activities such as cooking

Question: If salmon ceviche is on the menu – how is the salmon prepared?

- A: Marinated in red pepper and fried
- B: Marinated in lime
- C: Prepared in a plastic bag and put in hot water (60 degrees).

Question: In which of these dishes is there a lot of salt?

- A: Caviar
- B: Cheese
- C: Fruit salad

6. Avoid using negative expressions such as never, not etc.

Question: Which of these countries is not located in Europe?

- A: Spain
- B: Germany
- C: Syria

and school grades to promote motivation and inclusion. Table 20.1 shows some examples of questions and answers in the Quiz Tool.

More material can be added to the program by supplying more personalized information or questions of relevance to current school topics, and by giving feedback and comments related to the student's answers. The difficulty level of multiple-choice questions can be adapted to the student's cognitive level and knowledge.

The Quiz Tool can also be used for self-study, for example as an obligatory home assignment or a daily routine. It is possible to repeat the same quiz many times, and comments from parents and staff indicate that such repetitions may promote learning and feelings of mastery. The Quiz Tool may also be used for retrieval of materials that the student learned some time ago.

If possible, the quiz questions should be related to the mainstream curriculum. This goal will become difficult to achieve for students with JNCL as the disease progresses. A moderate or severe dementia requires that the quiz is modified to be appropriate for the individual person. A person-centered quiz requires good knowledge of the person with JNCL and his or her life. In any case, constructing a quiz based on the person's life, desires and experiences, requires a close collaboration with parents.

In the present project, some young persons with JNCL often selected the last presented alternative when given three alternatives in the Quiz Tool (see Figure 20.2). The decline in working memory capacity may have made it difficult

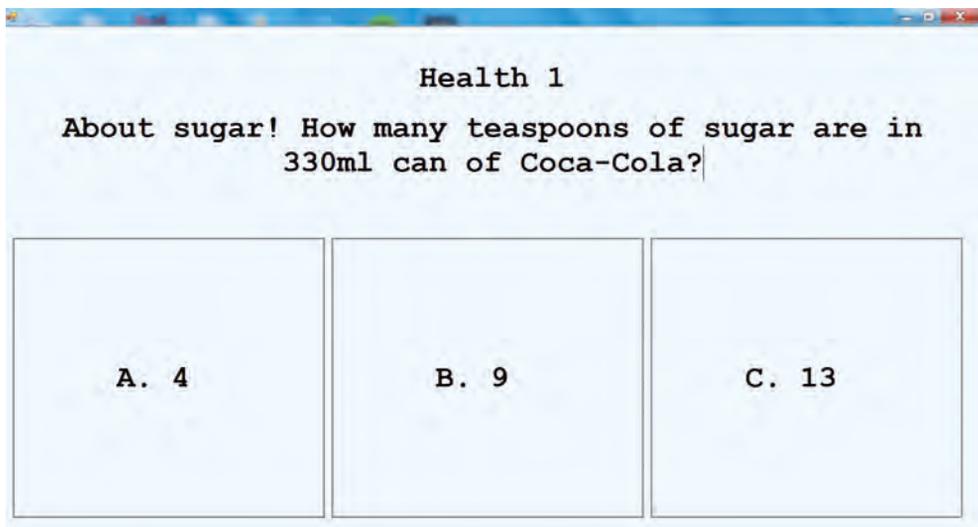


Figure 20.2 The Quiz Tool – a quiz question with three possible answers

The question and the answers can be repeated as many times as the player wants.

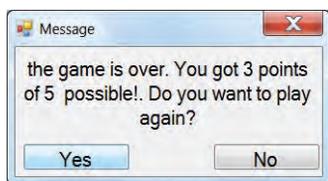


Figure 20.3 The Quiz Tool – end of the game

At the end of the game, the number of points is displayed and the player is asked if he wants to do the same quiz again. The information is presented in print, braille and by speech synthesis.

for them to remember all three alternatives. In spite of such difficulties, the quiz organizers (parents and staff) were never in doubt about the usefulness of the program. They said that the persons with JNCL usually showed great enjoyment when playing quizzes, just as they had done when they were younger. Playing quiz games raised their engagement and mood, and according to staff and parents, they were also more awake. Participating in enjoyable activities is an important element for quality of life and playing quiz games seems to provide a good social setting. Ensuring that some of the last quiz response alternatives are correct will promote feelings of mastery and pride, according to the parents and staff. At the end of each quiz, the players are asked if they want to do the same quiz again or leave it (Figure 20.3).

Observations indicate that playing quiz, in addition to supporting cognitive functions, also may be used to promote social learning in children and young people with JNCL. A specially constructed quiz can be a helpful element in handling or discussing difficult topics and situations, such as bullying, violence, democracy, or children's rights. It is not known to what extent quizzes are used to support teaching of such concepts, but they are an integrated part of the mainstream curriculum in many countries, and should also be taught to children and young people with JNCL. Maybe a tailored quiz about society and social behavior can contribute towards a better understanding of the importance of appropriate behavior for students with JNCL. Some teachers reported that some of the questions they included in the Quiz Tool had inspired the class, including the students with JNCL, to show good sportsmanship.

The Quiz Tool is available in English and Norwegian and can easily be translated into other languages.

Self-made quizzes

The Quiz Tool may be part of the teaching repertoire but it can also be used by children and young people with JNCL to construct their own quizzes, with their own choice of topics, questions and multiple-choice answers. There are some automatic quiz generators on the Internet but they are based on factual information found at Internet, and quizzes generated from them are therefore less

tailored to the needs of individual students, which is a necessity for young persons with JNCL (see Samuelsen, 2016).

It is possible to construct a quiz with or without the ability to see, but basic knowledge about operating the Quiz Tool is necessary. Reports from the present study indicate that a few young persons with JNCL were deeply engaged in creating their own quizzes and presenting them to peers and adults when ready. The parents further reported that building and creating quizzes resulted in strong positive feelings of ownership. The process of making their own quizzes stimulated the children and young people to investigate particular knowledge domains, such as sports, music or film. Parents and staff said that these investigations increased interest and motivation, which in turn lead to further study of the topic or domain. Self-made quizzes are good examples of self-driven education for these students. Scoring and announcing the results of the quizzes when they were played, and dispersing the prizes, were very exciting experiences for the students with JNCL.

It is possible to create a bank of self-made quizzes for later use in school or when meeting friends after hours. Self-made quizzes can also be shared with other students or friends through internet or email. The present study learned about a young man with JNCL who was very devoted to developing, sharing, and exchanging quizzes with others.

The Quiz Tool and collaboration

Observations from teachers in the Quiz Tool project indicate that doing quizzes in the class or in a group will motivate learners with JNCL to participate, be active and collaborate with peers, rather than working alone. Many students with JNCL preferred to work in teams and discuss solutions to quiz questions before answering. All the students in the class seemed inspired by the quizzes and tried to achieve a common group response in a positive and inclusive way. In some cases the questions and the answer alternatives elicited long discussions, and the exchange of ideas between the students provided some golden moments of implicit and incidental learning. The use of the Quiz Tool in the classroom thus seemed to contribute positively to the teachers' efforts to build positive attitudes and teamwork in the class.

The special interests and quizzes of a young man with JNCL

Many of the students who used the Quiz Tool appeared highly motivated and clever despite their severe disabilities. A young man with JNCL was a big fan of the books about Harry Potter written by the British author Joanne "Jo" Rowling.

He had read all the books and had become an expert on Harry Potter. This great interest made him compose his own stories about Harry Potter in braille with incredible imagination. His publications attracted public attention and were exhibited in a Norwegian library. The young man learned to make his own quizzes using the Quiz Tool and became a very devoted user of the Quiz Tool. He has constructed and dispersed several quizzes based on Harry Potter books for friends and peers with and without vision. The young man is also an expert on the films and stories about Star Wars invented by the American author and director George Walton Lucas. For some time, he has been busy making quizzes based on the stories from Star Wars.

Conclusions

Quiz games are commonly used in education and for pleasure. They can utilize different modalities and may be adapted to players with different abilities and disabilities, including blindness and dementia. Research on dementia and experiences with elderly people indicate that quizzes may stimulate cognitive function and well-being. The findings of the present study, using the computer-based Quiz Tool, show that persons with JNCL can take part in quizzes both individually and in groups. The quiz seems to stimulate their motivation, learning and the maintenance of skills and knowledge. Quiz games make repetitions and over-learning possible if used in a strategic way within and beyond education.

The visual, tactile and auditory options of the Quiz Tool inspire collaboration and inclusion in mainstream settings because sighted students and students with visual impairment can play together on an equal footing. The Quiz Tool can further be used to assess the students' knowledge in different domains, and to find out whether the student is ready for the next level or rather may need more help in the zone of developmental maintenance (see Chapter 2). In this way it can help the teacher to collect information for evaluation purposes in a non-threatening manner. Observations further showed that the Quiz Tool can be used to steer the students' attention towards specific concepts or subjects, and to promote inclusion and pleasure in social settings.

References

- Adesope, O. O., Trevisan, D. A., & Sundararajan, N. (2017). Rethinking the use of tests: A meta-analysis of practice testing. *Review of Educational Research, 87*, 659–701.
- Agarwal, P. K., Karpicke, J. D., Kang, S. H. K., Roediger, H. L., & McDermott, K. B. (2008). Examining the testing effect with open- and closed-book tests. *Applied Cognitive Psychology, 22*, 861–876.

- Alzheimer's Society (2018). <https://www.alz.org/uk/dementia-alzheimers-uk.asp>
- Balota, D. A., Duchek, J. M., Sergent-Marshall, S. D., & Roediger, H. L. (2006). Does expanded retrieval produce benefits over equal-interval spacing? Explorations of spacing effects in healthy aging and early stage Alzheimer's disease. *Psychology and Aging, 21*, 19–31.
- Bontchev, B., & Vassileva, D. (2010). Modeling educational quizzes as board games. Proceedings of IADIS International Conference e-Society, Porto, Portugal, March 18–21, 2010 (pp. 20–26).
- Brusilovsky, P., Sosnovsky, S., & Shcherbinina, O. (2004). QuizGuide: Increasing the educational value of individualized self-assessment quizzes with adaptive navigation support. In M. Driscoll & T. Reeves (Eds.), *E-Learn: World Conference on E-Learning in Corporate, Government, Healthcare, and Higher Education* (pp. 1806–1813). Norfolk, VA: Association for the Advancement of Computing in Education.
- Chang, S. H. (2015). Memory strategies used by teachers. *Ohio Journal of Teacher Education, 29*, 5–19.
- Cotelli, M., Manenti, R., & Zanetti, O. (2012). Reminiscence therapy in dementia: A review. *Maturitas, 72*, 203–205.
- Elder Care (2016). <https://www.which.co.uk/elderly-care/housing-options/exercise-and-entertainment/451176-cognitive-exercise-and-mental-health>
- Fritz, C. O., Morris, P. E., Nolan, D., & Singleton, J. (2007). Expanding retrieval practice: An effective aid to preschool children's learning. *The Quarterly Journal of Experimental Psychology, 60*, 991–1004.
- Karpicke, J. D., & Aue, W. R. (2015). The testing effect is alive and well with complex materials. *Educational Psychology Review, 27*, 317–326.
- Karpicke, J. D., & Bauernschmidt, A. (2011). Spaced retrieval: Absolute spacing enhances learning regardless of relative spacing. *Journal of Experimental Psychology: Learning, Memory, and Cognition, 37*, 1250–1257.
- Karpicke, J. D., Butler, A. C., & Roediger, H. L. (2009). Metacognitive strategies in student learning: Do students practise retrieval when they study on their own? *Memory, 17*, 471–479.
- Dormann, C. (2016). Toward ludic gerontechnology: A review of games for dementia care. Proceeding of First Joint International Conference of the Digital Games Research Association (DiGRA) and the Foundations of Digital Games conferences (FDG), Dundee, Scotland, August 1–6, 2016.
- Livingston, G., Sommerlad, A., Orgeta, V., Costafreda, S. G., Huntley, J., Ames, D., Cooper, C., et al. (2017). Dementia prevention, intervention, and care. *The Lancet, 390* (10113), 2673–2734.
- McDermott, K. B., Agarwal, P. K., D'antonio, L., Roediger III, H. L., & McDaniel, M. A. (2014). Both multiple-choice and short-answer quizzes enhance later exam performance in middle and high school classes. *Journal of Experimental Psychology: Applied, 20*, 3–21.
- Melero, J., Hernández-Leo, D., & Blat, J. (2014). Teachers can be involved in the design of location-based learning games: The use of the puzzle metaphor. In S. Zvacek, M. T. Restivo, J. Uhomobni & M. Helfert (Eds.), *Proceedings of the Sixth International Conference on Computer Supported Education, Volume 3* (pp. 179–186). Setúbal, Portugal: Scitepress.
- Morey, C. C., & Cowan, N. (2018). Can we distinguish three maintenance processes in working memory? *Annals of the New York Academy of Sciences, 1424*, 45–51.
- Samuelsen, J. (2016). Automatic quiz generation for elderly people. Master thesis, Department of Information Science and Media Studies, University of Bergen, Norway.

- Subramaniam, P., & Woods, B. (2012). The impact of individual reminiscence therapy for people with dementia: systematic review. *Expert Review of Neurotherapeutics*, 12, 545–555.
- Uner, O., & Roediger H. L. (2018). The effect of question placement on learning from textbook chapters. *Journal of Applied Research in Memory and Cognition*, 7, 116–122.
- Van Gog, T., & Sweller, J. (2015). Not new, but nearly forgotten: The testing effect decreases or even disappears as the complexity of learning materials increases. *Educational Psychology Review*, 27, 247–264.
- Woods, B., Spector, A., Jones, C., Orrell, M., & Davies, S. (2005). Reminiscence therapy for dementia. *Cochrane Database of Systematic Reviews*, Issue 2, Art. CD001120.

21

Games

*Bengt Elmerskog, Mohammed Beghdadi, Svein Rokne,
Anna Christal, Deirdre McElrone, Judith D. Oxley
and June Cameron*

A game is a structured form of play for enjoyment and pleasure, but games may also stimulate learning and cognitive functioning. In education, games may give positive challenges, improve performance, or maintain skills and knowledge. Games can be competitive, like chess, or non-competitive, such as role play. There are group games and games that are played individually. Both individual and group games may be experienced as exciting and motivating. Leisure time activities include games of which there is a great variety.

Children with JNCL may participate in group games on par with their peers when the games are constructed or adapted to facilitate inclusion of players who are blind (see Ardito & Roberts, 2007; Tzvetkova-Arsova & Zappaterra, 2018). Consider a deck of cards. The individual card can be marked in a corner with braille characters or Moon letters (see Chapter 14) according to a system where the suit (Hearts, Spades, Clubs and Diamonds) is written with the corresponding letter and the denomination is similarly marked just underneath (with letters for Ace, King, Queen and Jacks, and a number for the other cards). Pre-marked cards may be obtained from a number of sources, including national organizations supporting people who are blind or have low vision, or from commercial vendors.

Dice are important elements of many games and can be constructed with embossed numbers to be read by people who are blind. Dice are used in Yahtzee and in board games like Ludo and Monopoly. Dice may serve as useful educational tools in addition to their primary use. Using dice provides cognitive exercising, particularly in games where two dice are used as in Monopoly or in Yahtzee where decisions must be made based on the face value of five or six dice at a throw. The tactile properties of texture and Velcro can be used to differentiate between game pieces. Attaching magnetic tape to boards and game pieces can reduce frustration arising from pieces being knocked out of position. The board games themselves can be marked in such a way that children and young people with JNCL may be

able to participate with little or no intervention from an assistant, at least initially. There are many resources readily available on the internet (e.g., <http://www.pathstoliteracy.org/strategies/tips-adapt-games-children-vision-impairments>).

Research indicates that intellectual and motor exercises might help people with dementia maintaining skills and abilities (see Chapter 5 and 7). Games provide mental stimulation and may function therapeutic and slow down mental decline in people with cognitive disorders, including playing strategic games, assembling puzzles, playing board games, chess or card games. Playing games may also support communication, literacy, and social interaction. Games that earlier were enjoyed by the young person with JNCL are usually a good starting point. Many games can be adapted to meet the changing needs of the young person as the decline in skills becomes more apparent. New games can also be introduced, but it is important to analyze the complexity and abilities needed.

Over time, it will gradually become more difficult for young people with JNCL to read the boards, the cards or the dice. Thus, there is a shift from *independent play* to *interdependent play* (see Chapter 16) when the degree of assistance when playing increases, but the feeling of participation (and indeed the gloating when winning) does not diminish even as these skills deteriorate. Verbal cueing and verbal orientating assistance become necessary and dialogue becomes a (pleasurable) part of the game.

Experiences of parents and staff

In the present project (see Appendix A), both parents and staff mentioned the use of games for education and leisure. A counselor and teacher expressed the following:

Throughout my 25 years of experience working with students who have JNCL I have experienced that games are very popular activities for students with JNCL. Playing games is stimulating their cognitive function and reducing anxiety, maintaining relationships leading to more enjoyable moments. I have also seen that students are concentrated, aware and conscious – playing games can actually help them retrieving memories.

Popular games need to be modified in line with the progression of different declines for individuals with JNCL. Such modifications may entail more structured settings, more support from fellow players, or making key information available through touch and hearing. Modifications of games may support further engagement for games, as described by these parents:

We modify the original games and adjust them to her mental progress. She plays very often but she has always preferred quizzes. Her interest in games grew over time.

The comments from parents and staff show that the amusement of participation in competitive activities is preserved throughout life for individuals with JNCL. A few parents have even reported that the children's interest for playing games grew with age. Winning games or the excitement of the possibilities of winning, is expressed by parents as a thrilling experience throughout life. The zest for competition somehow becomes a part of their child's personality, as described in the following two quotations:

He's really into the games on the iPad like Battleship and Plants Versus Zombies. He plays it and he talks about it whenever there is a listener.

He loves to play Sponge Bob and video games, but he loves to talk about it even more.

However, staff and parents see games as more than just an activity where one can win, lose or beat another person or group; ordinary daily activities like shopping, walking to school, or reading can be made into a game. The following teacher is using shopping to teach mathematics:

Learning about money and mathematics was done through playing shopping and handling money. We used reading games to learn how to use a magnifying glass and other technical aids.

The extent and quality of a child's engagement in an activity is a matter of how the activity is perceived by the child and how the exercise is presented to the child. An activity may appear boring unless steps are taken to make it more like a game that is enjoyable and meaningful to the child. Games may thus serve an additional purpose beyond the actual playing. Many parents and staff members emphasize that strategically adapting potentially "boring" activities into ones that have the properties of games is an effective way of enhancing learning and maintaining knowledge. Participating in activities attached to educational themes, objects or topics, was perceived as playing games by the participants with JNCL, sometimes resulting in astonishing and positive side effects on learning:

A calendar box was introduced to him to push the button to communicate when he wanted to eat or read, but he looked at it as more of a game. He learned braille by doing this. The same with the Go-Talk – a speech augmentation device – he learned how to use it through playing.

She learned a lot about nature via the game «Der Natur auf der Spur» («On the trail of nature»). It was about nature, it was very informative. The game included pictures of trees and various questions and so on and she learned it all. But it was not her hobby. She preferred reading and listening to music or to cassettes.

We thought this game would be done only once, not a long-term enjoyable activity. We play with a large collection of porcelain cats. Our daughter's role was to name the cats, touch and explore them, to describe their size, length, thickness, type of tail, placement of the ears, and so on. She always asks for this exercise or game. I can even ask her «What colors do the cats have?» and she can manage doing that as well. It is remarkable and very pleasing that she is able to devote herself to these cats for such a long period of time. This game is good memory training, my daughter is not aware that I'm actually working with her, and that's the wonderful thing about it.

Games have been used to achieve different kinds of skills and knowledge like in this case:

In the game "Performing magic" something is placed under a cloth and our son must say what it is by exploring it in a tactile manner. For instance, he learned the difference between metal and plastic by doing this. Similar methods were used to distinguish between different sounds and so on. We do all these things and they may not only work well, but he has a lot of fun doing such exercises.

A teacher of children with JNCL emphasizes the use of quiz games to achieve learning:

Quiz is just a wonderful way of combining pleasure with learning. «What's the capital of ... and which river passes through ...?» Then they answered and were very proud and happy. That was helpful. Or we utilized their knowledge of foreign languages: «Which language do you speak?» or «What is good morning in French?» and so on, that's all possible if it accords with the interest or knowledge of the kids.

The use of Quiz is described further in Chapter 20.

Some parents emphasized the potential of using games to achieve learning and participation in schools. One parent mentioned games from Royal National Institute of Blind People:

There are some games in the school like dominos and different RNIB games marked with braille. I'd like to think [laughs] that there might be someone that could play a game with him, but it does not happen, you know – adults could facilitate but – I don't know. I don't think they understand the potential in using games as a motivator for learning and socializing.

An educational counselor expressed the following:

I have experienced that the disease can cause, unfortunately, individuals with JNCL to withdraw from everyday relationships and activities with friends and family. Playing games is one of many ways of maintaining and building up such relationships.

Modifications and engagement

The visual impairment is the first of several barriers to participating in games. Most games require vision. There are games adapted to the needs of people with blindness and sources of ideas for how to modify them or for commercially available adapted equivalents, but they are not generally known. It is always a danger that declines will lead to non-participation in playing games if not met by appropriate measures like in this case:

She liked playing cards – she knew how to play cards. But it was difficult when she got blind. She would have been interested in it. This interest was replaced by listening to audio books.

The example illustrates many dimensions of the compounding problem: loss of participation opportunities may lead to losses in other domains, such as socialization, communication, emotional engagement and expression, and so forth. Although listening to audio books can be pleasurable and educational, and may provide topics to discuss with others, it becomes a passive activity if not shared with others. Passivity has been noted as a concern in adolescents with JNCL (see Chapter 4).

Participation opportunities in play are also lost because of other factors than deterioration of vision. The decline in communication and onset of childhood dementia may make situations with game playing rather complex, in particular when the child or young person has problems in remembering what happened last or when the overview of the playing activities is lost. Some parents and teachers have the experience that participating in one-to-one games is easier for individuals with JNCL when communication problems and childhood dementia become more apparent. Moreover, many parents and teachers emphasize emotional engagement by those participating as a key factor to achieving pleasure and entertainment in activities:

Playing games works when people enjoy the game as well... yeah. For him at the moment the thing that engages him most is his physiotherapy session. He's most alert and engaged, he almost considers the physio as a game to play. I know we just felt really strongly that they (the helpers) needed to be engaged, you know, physically and mentally engaged, no matter where they were at.

These parents' comments emphasize the fact that support is more than practical help; in fact, the staff's emotional engagement may be decisive for how a person with JNCL functions in the zone of developmental maintenance (see Chapter 2).

A variety of games

Many games are already accessible either because of their inherent properties, or because there are commercial versions appearing on the market. It is possible to make existing activities and products accessible to children and young people who are blind by adapting materials and rules (e.g., the complexity of a game) so that participation continues to be possible even in the face of declining vision and cognition. The following sections provide general ideas about the kinds of adaptations that are possible, together with explanations of how these modifications may support ongoing participation in game activities.

There is a rapidly increasing pool of online resources that can provide more ideas, and greater detail about how to adapt specific materials, where to obtain ready-made products, and how to integrate the materials and activities into activities of daily living, education, and leisure. Many popular games are now available as physical packages or as apps for mobile phones, tablets, and other computerized media. Large screen television and computer monitors, particularly

those with touch screens, can increase the size and luminance of popular games, rendering them more accessible than they would be if presented on a tiny screen.

Online searches can include key words for games for specific populations (e.g., people with dementia, intellectual impairment, or visual impairment) in addition to games according to skill sets (e.g., strategy, memory, words, numbers), and the medium (real or virtual on an app). National support organizations maintain extensive online lists of resources and can serve as good starting points for searches.

Board games

Creating words in board games may promote learning in reading and writing, and such games are available for persons with visual impairment. *Scrabble*® (Hasbro) can be shared with sighted peers. Scrabble with braille works best for advanced braille users in collaboration with sighted helpers. It is possible to purchase versions equipped with everything needed for individuals with JNCL: braille tiles, tactile game board and braille and audio instructions. The game board is pasted



Figure 21.1 Scrabble with braille

onto a wood backing and covered with a clear hard plastic film that is ridged along each cell and acts as frames in which a player places a tile. Each cell has printed information and letter tiles with raised braille markings.

The *Scrabble* word game shown in Figure 21.1 is rather advanced. There are simpler versions of word games, such as the *Bananagrams*©. The *Bananagrams* set consists of 144 pieces in braille and print on a white background. It is easier to master because the young person with JNCL can form words in collaboration with family and friends. The words are readable for players with full sight, low vision and blindness. The manufacturer supplies a version for people with low vision (*Bananagrams Big Letter*).

Choice Words© (MindWare) is a rather simple but funny game that involves auditory recall by asking students to identify words, terms, titles, and phrases that involve the root word. For example, the root word *cut* may stimulate recall of phrases that include it, such as *cut rate, cut throat, haircut, paper cut, cold cut, short cut, cutting edge, cutting board, cutting room floor, cut-and-paste, cut-and-run, or cut it out!*

There is a commercial braille version of *Monopoly*© with dice and cards in large print fonts and braille (*Braille and Low Vision Monopoly*).

Blindfold Sudoku is for players with sight and visual impairment and available as an app (iOS). It comes with five Sudoku 9-by-9 puzzles – easy, medium and difficult – for audio play and is controlled through iPhone gestures. Students solve the puzzle by listening and imagining the puzzle layout in their mind.

A tactile-based adaptation of *Nine Men's Morris*, a traditional strategy game, consists of a grid with twenty-four intersections or points. Each player has nine pieces or "men" that usually are colored black or white. Players try to form "mills" – three of their own men lined horizontally or vertically – which allow a player to remove a man from the opponent. A player wins by reducing the opponent to two pieces (when it is no longer possible to form a mill and thus winning is impossible), or by leaving the opponent without a legal move.

Follow your Nose is a game that encourages exploration and discovery through the sense of smell. It stimulates both the olfactory sense and cognition while it elicits reminiscences. There are about 30 distinct aroma diffusers in tamper-resistant flasks which the players match to the corresponding images. The images must be adapted to tactile or auditory use to make the game accessible for persons who are blind.

Matching games

In these games the players match an object to a word in print or braille, a photo card or similar. Good games to play may include guessing games with a box, sack or tray containing familiar objects that the players must identify. These may

include everyday objects which can be named, classified or sorted. This is a useful game to support key vocabulary. Figure 21.2 is an example of a matching game.

Figure 21.3 shows a game where the players are matching a card with a braille or printed word to a corresponding object. Games that stimulate mental capacity through word-related activities, such as word identification and odd one out, help support and maintain word identification and memory.

Dominoes is a family of matching games that easily can be modified for players with low vision, blindness and cognitive disorders (see Figure 21.4). The dominoes can be played with the tiles placed face-down and mixed. Each player selects seven domino tiles which are placed in front of the player but hidden from the opponents. The remaining tiles constitute a drawing pile. The player with the highest double (six-six) starts the game by placing the double on the table. During each turn, the player selects a domino tile from his pile that matches at least one side of one of the domino tiles on the table. A new domino tile is picked from the draw pile if none of the players'



Figure 21.2 Game tray with matching pairs



Figure 21.3 Game where words are matched with corresponding objects

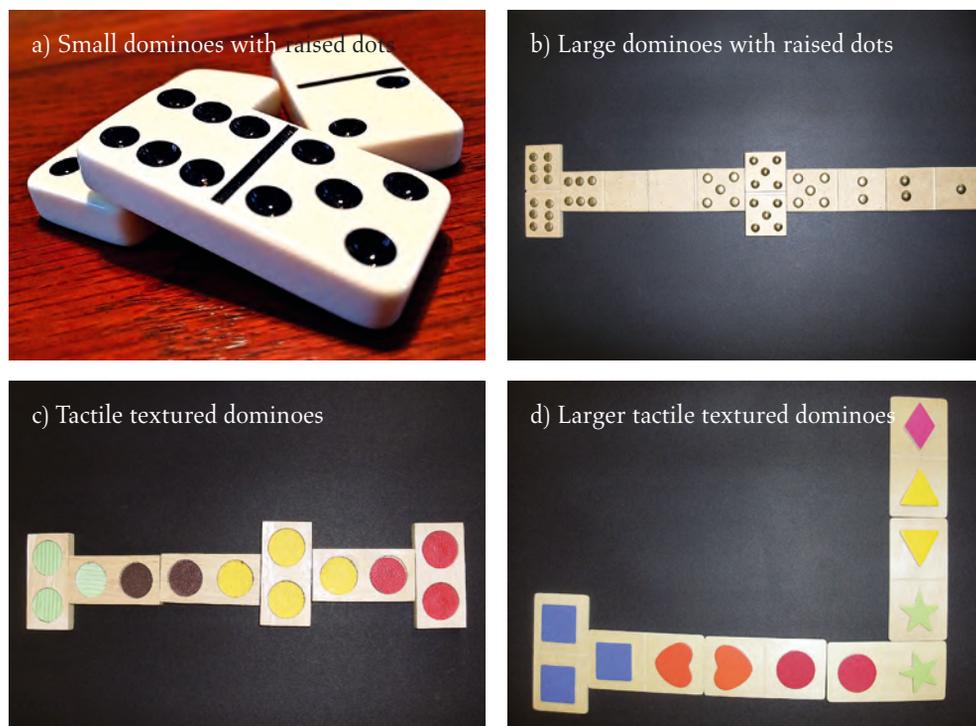


Figure 21.4 Dominoes with four different forms of adaptations

dominos can be played. When the draw pile has been depleted, the current player must pass without making a move. The first to place all the tiles is the winner. Playing Dominoes may support the maintenance of number recognition, counting and many other cognitive skills, as well as competitiveness and the motivation to win. Versions designed for people with blindness and low vision are commercially available.

Memory games

Memory games may promote concentration and memory skills. There are many memory games such as apps, tactile games and digital games.

The Sarepta digital memory lotto game (Sarepta Studio, Norway) is a tactile and audio version of the classic memory card game (see also Chapter 19). The purpose of the game is to challenge the memory by linking corresponding sounds, words, pictures and so forth in pairs in categories like animals, nature, transport, humans or geography. It can be used to make the player familiar with areas of special importance or interest.

The *Audio Memory Game* in Sarepta uses a speech synthesizer. There are 16 hidden mixed audio cards numbered from 1 to 16. There are eight identical pairs within the 16 cards, and the players have to find two matching cards. The game will continue to the next player if a player makes an incorrect matching of cards. The players navigate from card to card by pressing a switch, the speech synthesizer will identify each card, for instance by saying «*card number one*» followed by a sound from a *dog barking*. A similar card with a dog barking can be found from for example card number eight. It is important for the student to listen for clues and remember them when other players are doing their trials. The game is over when all the cards have been matched, and the winner is the player with the most matches. The Audio Memory game can be played individually or as a group activity.

The purpose of *Shake Loose a Memory* is to recall memories and share emotional events with others (reminiscing). The game can provide social interactions and stimulate to meaningful discussions. The game consists of a single die and cards. Participants roll the die and receive a corresponding card. Each card starts with a simple instruction, like *keep this card if you have been a scout*. If yes, the student will respond to a question on the card, such as *remember earning any badges?* If the answer is no, the player will be asked to roll the die again or the game will continue to the next player. This game is not about *winning* or providing the *best* answer. It is about maintaining memories and sharing the personal history and good memories with others, in many ways imitating an ordinary conversation. An important consideration with games of this type is the importance of having a group of people playing together who actually share memories and are able to co-construct, and as cognition declines, this consideration assumes greater significance. The game can be adapted to very local, very immediate experiences (e.g., events at school this month, shared TV viewing, shared sport event participation, family activities this month) to accommodate memories. This and similar games can contribute to making classmates more acquainted with the student's home situation and leisure interests, or the family more acquainted with the student's situation at school.

Strategy games

These games require planning and finding solutions, and there are many old favorites, such as *Monopoly* and its numerous spin-off variations based on different themes and places. A quick online search will locate many versions.

Many children find *Connect 4* exciting and entertaining. This game is played by two players and may be played by individuals with low vision or blindness. Each player is given their own plastic discs that can be identified by color or

tactile recognizable patterns, for example black Velcro stickers. The game is about dropping colored discs from the top into a seven-column, six-row vertically suspended grid. The pieces fall straight down, occupying the next available space within the column. The objective of the game is to connect four discs of the same color or tactile pattern next to each other vertically, horizontally, or diagonally before the opponent (see Figure 21.5). The game may contribute to maintaining number and counting skills, tactile discrimination and memory, and the most important goal of all – to have fun. At the simplest level, the game can entail no more than turn-taking between participants in dropping in the discs and enjoying the resulting noise. Even this level of the game promotes active participation and socialization by simplifying the "rules" and thereby making it fun. It is possible to purchase large Connect 4 games for indoor or outdoor use. The large versions are easier to manipulate if the player has problems with fine motor skills. A small hole may be drilled into one set of counters or discs to aid identification.

Pass the bomb is a thrilling game (Figure 21.6). The players sit in a circle while the bomb is passed around the group. The players are asked to think and express a word connected to the *Theme of the Day* when holding the bomb.



Figure 21.5 *Connect 4* with black Velcro for tactile discrimination, red is the winner!

Themes can be things that are found in a supermarket, classroom, bedroom, or related to a special event, for example Christmas or summer holiday. The aim is to not be holding the bomb when it surprisingly goes BOOM! This game may stimulate memory through word-related activities and word identification. It helps improve the ability to recall by encouraging the recollection of memories via verbal interactions or pictures. Old fashioned versions include *hot potato*: no bombs go off, but there is plenty of noise when the player left holding the hot potato screams in dismay!

Noughts and Crosses (also known as Tic-Tac-Toe) is a strategy game that can be composed of a wooden 3D set (see Figure 21.7), or it can be accessed as an app for mobile devices. The players take turns to place their X or O into one of the empty squares in the grid by clicking on it. To win the game, the players must get three of their own symbols in a line horizontally, vertically or diagonally. Noughts and Crosses may help maintaining memory skills, tactile discrimination, strategy use and problem solving. It may also support the maintenance of number and counting skills and fine motor skills. The competitive element appears to enhance the players' enjoyment of the game.



Figure 21.6 The bomb card game

Shut the box is played using dice and a box that has nine levers on hinges, each marked with a numeral and there are one-player and two-player versions (see Figure 21.8). The object of *Shut the box* is to shut all nine levels. One player rolls both dice onto the green baize floor of the box, and shuts the lever(s) according to a number or numbers face up on the rolled dice. If a player reaches a position where the number rolled has already been used to close the corresponding lever, the play passes to the second player. At the start of the game, all levers or tiles are open, showing the numerals 1 to 9. Each player plays by throwing or rolling both dice into the baize. The player adds up the dots on the dice and then "shuts" (closes, covers) one of any combination of open numbers that equals the total number of dots showing on the dice. For example, if the total number of dots is 8, the player may choose any of the following sets of numbers (provided that all the numbers in the set are available to be covered): 8, 7+1, 6+2, 5+3, 5+2+1 and 4+3+1. The player



Figure 21.7 Noughts and Crosses (Tic-Tac-Toe)



Figure 21.8 Shut the box

then rolls the dice again, aiming to shut more numbers. The player continues throwing the dice and shutting numbers until reaching a point at which, given the results produced by the dice, the player cannot shut any more numbers. At that point, the player scores the sum of the numbers that are still uncovered. For example, if the numbers 2, 3, and 5 are still open when the player throws a one, the player's score is 10. Play then passes to the next player. After every player has taken a turn, the player with the lowest score wins. If a player succeeds in closing all the numbers, he or she is said to have "Shut the Box" and wins immediately, and thus the game is over.

In *Build a Beetle* players take turns throwing a die and cannot begin building a beetle until they have thrown a six, which allows them to take a beetle's body (Figure 21.9). A larger tactile die

may be introduced as and when the regression of skills becomes apparent. When a six is thrown, the player has a second turn, and this rule applies throughout the rest of the game whenever a player throws a six. Once the body has been taken, the subsequent parts are selected when the appropriate die value is thrown, as follows: 5 gives the neck, 4 gives the head, 3 gives the tail, and 2 and 1 give a leg. *Build a Beetle* provides tactile stimulation and stimulates number recognition, counting, communication and memory, and helps maintain fine motor skills. The game is competitive and many young people with JNCL find it enjoyable.



Figure 21.9 The Beetle Game with an ordinary and a tactile die

Construction games

Most construction games can be played individually or in collaboration with others. *Meccano*, *Lego* and *Duplo* are examples of constructions games where children and young people are stimulated to construct vehicles, bridges, helicopters or cranes. Many of these games promote children's interests in areas like science, technology, engineering and mathematics.

There are many construction games suitable for individuals without sight. Many structures in the surroundings cannot be perceived as instant holistic features by children who are blind. Structures may be easier to conceptualize through the use of construction games. For instance, building a bridge may make it easier to perceive or understand the structure of a bridge when vision is no longer available. Construction games can be used strategically in education by enabling children and young people with JNCL to act as constructors.

Construction games promote fine motor skills and incorporate different learning styles, such as learning by doing. Learning about physical structures and to imagine them and form a holistic representation without vision might be difficult. Such challenges can be compensated by organizing settings where the child becomes the constructor of a structure (e.g., a spaceship) or a part of the

structure (various projections from the spaceship). Providing the necessary support for the child's construction might facilitate the ability to form an overall concept or idea about the structure based on both earlier visual experiences and present tactile knowledge. By creating different kinds of space craft, the student can build up conceptual knowledge at a categorical level that serves as an overall template, together with the various optional features that might create subcategories. In typical development children learn to construct houses, barns, skyscrapers, and so forth. In so doing, they discover natural categories of "buildings" and elements that may be used to create specific structures.

The picture in Figure 21.10, sent by a parent, shows a young boy with JNCL building a space shuttle with Lego. Lego building has been one of his hobbies for a long time and his skills in mastering Lego are astonishing. The parents and his teacher believe the tactile skills developed through playing with Lego has been a vital asset supporting the boy's performance in reading and writing braille.

Construction games can be used for preparing the child to learn skills within reading, writing and mathematics and the understanding of objectives and

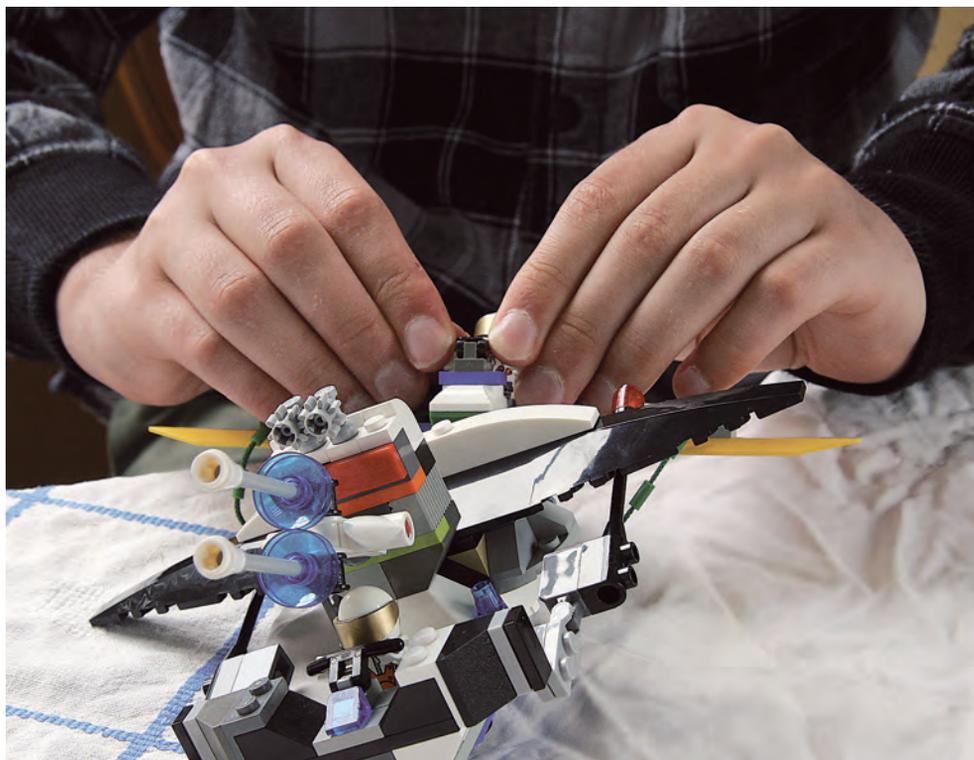


Figure 21.10 A boy with JNCL constructing with his Lego

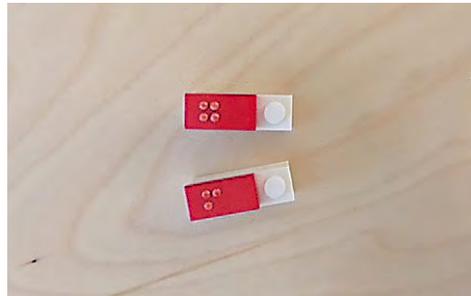
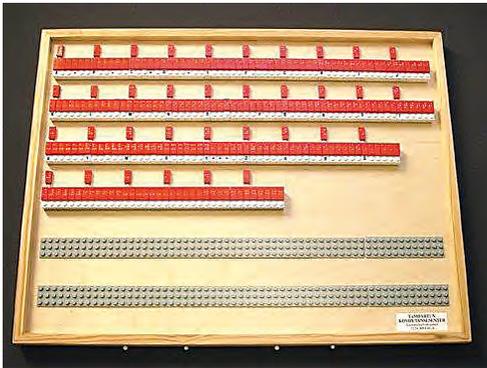


Figure 21.11 Lego braille frame and two examples of bricks with braille letters

structures in the society that otherwise cannot be perceived by other means than through vision. Lego and other construction games have been used as motivators to achieve reading and writing skills in braille. Lego bricks have also been used to teach letters and construct words and sentences (see Figure 21.11). Construction games may support the understanding of shapes and spatial relations, as well as number recognition and counting.

Other games

There are many other games available for young people with visual impairment, like *Chess*, *Othello*, *Parcheesi*, *Checkers*, *Chinese Checkers*, *Racing Crown*, *Tactile Backgammon*, *Snakes and Ladders*, *Rubik's cube with touch marks*, *Sudoku in color and braille* and *Tic-Tac-Toe*, *Slim*. Note that many sports games can be adapted as well (Ardito & Roberts, 2007).

Conclusion

Games are a source of enjoyment and learning. Playing games is important for all human beings, particularly children and youth. Playing games encompasses personal entertainment, socialization and enjoyment – important aspects of the quality of life. This short review shows that it is possible to adapt games of all types to enable children and young people in different phases of JNCL to participate actively and with enjoyment, and how participation can support both development and maintenance of many skills. Games provide mental stimulation and may be considered as therapeutic for people with cognitive disorders. Playing games may in fact contribute to delaying the decline of functions in the zone of developmental maintenance.

In sum, playing games should be an option for most young people with JNCL and might be used as an effective method for achieving learning and preservation of skills in education. An added benefit of playing games in groups is its contribution to building social affiliation and a group identity. Being in a competitive mode may contribute to increased motivation for learning and may enhance patience and skills during performance.

References

- Ardito, M., & Roberts, J. (2007). A catalog of physical activities for visually impaired youth. Bachelor thesis, Worcester Polytechnic Institute
- Celeste, M. (2006). Play behaviors and social interactions of a child who is blind: In theory and practice. *Journal of Visual Impairment and Blindness*, 100, 75–90.
- Giannakopoulos, G., Tatlas, N. A., Giannakopoulos, V., Floros, A., & Katsoulis, P. (2018). Accessible electronic games for blind children and young people. *British Journal of Educational Technology*, 49, 608–619.
- Neugnot-Cerioli, M., Gagner, C., & Beauchamp, M. H. (2015). The use of games in paediatric cognitive intervention: A systematic review. *International Journal of Physical Medicine and Rehabilitation*, 3 (4).
- Rettig, M. (1994). The play of young children with visual impairments: Characteristics and interventions. *Journal of Visual Impairment and Blindness*, 88, 410–420.
- Tzvetkova-Arsova M., & Zappaterra, T. (2017). Play in children with visual impairments. In S. Besio, D. Bulgarelli & V. Stancheva-Popkostadinova (Eds.), *Play development in children with disabilities* (pp. 181–200). Berlin: De Gruyter.

The Social Life of Children and Adolescents with Juvenile Neuronal Ceroid Lipofuscinosis: School and Participation

*Stephen von Tetzchner, Anne Margrethe Andersen,
Anne Hagedorn Hamann and Susan Fugger*

Social relations and participation are core elements in human life. Social participation includes family, peers and the community. Parents and siblings are usually children's first social relations; they have significant influences on the children's development and continue to remain important in adulthood. The mutual influences between children and parents are important but children also engage in interactions with peers in events and activities inside and outside of school and kindergarten. Peers have a prominent place in children's social lives from an early age, and with age peer relations become gradually more important (Chen, French, & Schneider, 2006; Rubin, Bukowski, & Bowker, 2015). Observing and interacting with peers help younger children learn collaboration, conflict management, moral reasoning and other social skills. Among older children and adolescents, peer relations also contribute to the development of identity and a broader understanding of emotions and thinking in other people.

This chapter is about the development and maintenance of peer relations and social participation of children and young people with juvenile neuronal ceroid lipofuscinosis (JNCL). It is a common finding that disabilities will influence children's interaction with peers (Schneider, 2016). The declines in vision, cognition and motor skills of individuals with JNCL will influence their friendships and peer relations but the present study (Appendix A) shows a varied pattern of involvement in peer interactions in this group.

Vertical and horizontal relationships

Children and adolescents engage in vertical and horizontal relationships. In vertical relations, one part has more knowledge and social power than the other, and the relationship is asymmetrical and complementary. Adult-child relationships are typically vertical. Adults have control and children are given protection and

security. Children seek knowledge and help, and adults provide it. Peer relations are usually horizontal; the social power is more equally distributed, although children may have different roles when interacting. One throws the ball, the other catches. One dresses the doll, the other pushes the cart. Peer relations are important because they offer experiences and learning that vertical relationships cannot provide, including both competition and cooperation (Bukowski & Adams, 2005; Hartup, 1999).

Sibling relationships are usually based on some sense of being equals, even though differences in age, development and physical strength may imply differences in status. When one sibling has a disability, this is likely to change the relationship in some ways, and it may become more vertical than the age difference indicates. The relationship is basically the same as for siblings without disabilities but it is characterized by more warmth and care than other sibling relationships (Meltzer & Kramer, 2016; see Chapter 25).

Friendship is a horizontal relationship characterized by equality, symmetry and a strong emotional bond. Friendships may have positive influences on children's development: Friends develop social skills and understanding together, talk about themselves and others, and gain insight into moral values, conflict resolution and social and cultural rules (Bagwell & Schmidt, 2011; Dunn, 2004). Children who have friends generally show better development, adaptation and performance at school than children without friends. Friends have more fun together, often engage in more advanced forms of play, and solve school assignments better together than nonfriends (Bukowski, Motzoi, & Meyer, 2009; Hartup & Stevens, 1997; Laursen & Pursell, 2009).

Activities, peer relations and JNCL

Disabilities may influence a child's activities and losing acquired skills may be experienced as dramatic. A negative impact of reduced ability to manage everyday activities has also been found in elderly people with dementia (Barca, Engedal, Laks, & Selbæk, 2011).

Equally important, the loss of skills may influence friendships and other peer relations. Children with visual impairments may be excluded from many activities and tend to have fewer friends and smaller social networks than children without such impairments (Salminen & Karhula, 2014). There are many friendships between children with and without a disability but the prevalence of true reciprocal friendships involving children with severe disabilities is lower than friendships among children with typical development. Peer interaction may depend on adult intervention and "friends" with typical development may sometimes act more as

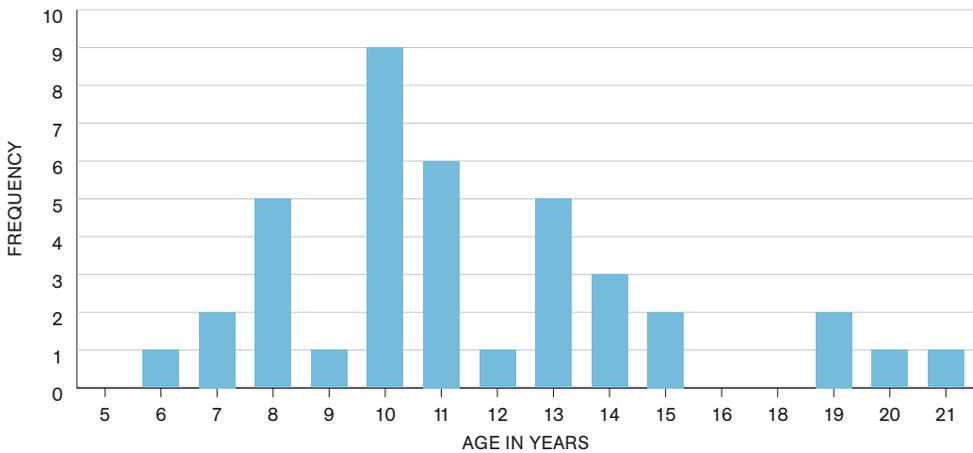


Figure 22.1 Frequency of first observation of loneliness at different ages

helpers than as friends (Guralnick, Neville, Hammond, & Connor, 2007; Rossetti & Keenan, 2018; Sterrett, Shire, & Kasari, 2017).

For children and young people, the school is the most important social environment outside the home. Education and the educational setting are strong contributors to children's social and emotional development and sense of identity and place in the world (Abdi & Guo, 2008). Children who are blind may attend a mainstream school but find interaction with peers difficult (Opie & Southcott, 2018). Also, intellectual disability tends to make social interaction with peers difficult and to increase the probability of loneliness (Gilmore & Cuskelly, 2014). In the present survey (Appendix A), the first signs of loneliness often appeared in late childhood and early adolescence, but sometimes earlier or in late adolescence or early adulthood (Figure 22.1).

For students with JNCL, the school and classroom may be the only sources of social interaction outside the family, and as the disease progresses social interaction in school may also become limited.

Well, she doesn't really have a social life. To be fair she doesn't have any friends as such outside school. She has got a supportive boyfriend; we do see him on a regular basis, every week.

Because of his disorders he lived at home isolated from other children. The kids didn't want to play with him any longer. From age eight he lived in a residential home during the week and in this way he was in touch with kids having the same problems. They were all the same.

How students with JNCL prosper and function at school will depend on the social environment, the relationships with other students and the degree of adaptive education. Loneliness is inversely related to acceptance and mutuality in the peer social environment (Newson, Mallow, & Watson, 2015). Loneliness is more frequent in young people who are blind, and a lack of mutual friendships will lead to loneliness (Gold, Shaw, & Wolffe, 2010). This quotation is from a young boy with JNCL.

I have become one of the world's most lonely persons, I don't have any friends, and my earlier friends stopped visiting me after I became blind.

Shared activities form the basis of early friendships. When preschoolers with typical development are asked about their friends, they answer that they are friends because they do things together. Participation in joint activities is an important criterion for friendship among older children and adolescents as well. With age, however, perceptions of friendship increasingly involve feelings of perceived closeness and shared emotions. Friends are understanding, loyal and can be trusted. Older children associate friendship to a greater extent with having similar attitudes and values; yet their friendships continue to be based largely on shared enjoyment of the same activities and the same music. In adolescence, descriptions of friendships additionally begin to include shared secrets and feelings, and opportunities for emotional support (Bukowski et al., 2009; Mathur & Berndt, 2006).

In the early years, children with JNCL engage in social interaction in the same ways as their peers, based on joint activities, but quite soon the visual decline makes participation in many activities involving gross motor skills difficult.

That is something that we have seen declining over the last year and a half when she started here in fifth grade. We had some issues last year and it's continuing to be an issue. She does not have very good relations with a lot of her peers.

Childhood is characterized by a high degree of gender-specific activities. Boys participate more in sports and group activities, while girls more often collect in smaller groups and are more preoccupied with intimacy and who is in and outside the clique (Maccoby, 1990). This may also influence the peer interactions of children with JNCL.

His loss of vision made participation in favorite activities like football in the break impossible, and because of this he also lost interaction with his peers. Some of the girls wanted to be with him but he usually ended up in a group with two or three girls, he talked about other things than they were interested in. He was interested in fantasy stories with dramatic events, spaceships and wars, and the girls talked about their own things and only half-heartedly engaged in his play.

Change of school may function well from a narrow educational perspective but may be more problematic for social participation and relationships because it is difficult for a young person with JNCL to make new friends. It is a gradual process, some maintain friendships longer, some lose them in transitions, and this should be a topic when transitions are planned.

The loss of sight was the reason for the change of school. The loss of friends followed. After that the increasing loss of skills made him restless. He became more isolated. In some stage of restlessness it was not possible to have him in the classroom any longer. On the other hand he enjoyed to stay with people, especially when they were in a good mood, when there was action and noise.

It is important to emphasize that the loss of friends is usually not absolute and a few regular friends may be sufficient for a good social life.

Her social life at present is amazing. There are 12 girls on this unit; we have got a boy unit upstairs and a girl unit downstairs, and a mixed unit with youngsters in chairs. She sees and hears them every well five days, four nights a week Monday to Friday. We socialize quite a lot together for birthday parties and other events, as I say going out to the theatre, it's not just the girls, and it's the boys and downstairs. She meets outsiders who come in; we've got young people as volunteers.

John has always had a good social life. And I would say that action and having a social life always made him smile and he loved taking part. He always enjoyed going out on visits or people visiting him. Going to places. And he would also be well visited by the familiar people. Familiar people are always very important but he also liked meeting new people. But he did have a little glint in his eye too as well because he would sometimes see how far he could go too as well in maybe trying a chat up line or something like that.

People with dementia are typically very dependent on other people (Wogn-Henriksen, 2012). Perhaps the most difficult thing about having dementia is the consequences the disease inflicts on the quality of being together with others, not least the closest ones. If a person cannot communicate verbally as well as before, the roles in the family and the relationships with friends change. For everyone, whether they have dementia or not, maintaining close and open contact with those who matter most to them is important for quality of life, as well as security and stability in daily living. The cognitive decline may manifest in many ways. With age, the activity repertoires of students with JNCL gradually get smaller, and their interests diverge from those of peers. As a result, they may be perceived as less interesting communication and interaction partners. Several parents describe this situation.

Peers usually approach and play with Thomas for a limited time, then lose interest when he can't engage in their games. Staff will sometimes endeavor to lead and steer games that can include all but these games are hard to sustain.

It is more and more difficult for him to be with his class. They are growing from him, have other interests and find it difficult to talk with him.

Because interests and activities are an important basis for friendship and peer relations, the interests of children or young persons will also influence their social interaction and participation in general.

His social life really is driven by how he is at the moment, which is that he gets very frustrated and he gets very anxious. So his social life at the moment is just very restricted to being in the lounge with the other young people. Listening to movies. It is very much driven by him at the moment. He does seem to prefer just to be quiet, knowing what's around him and understanding what's around him.

It is not only the range of the person's activities that may be limited, but also the type; some students with JNCL develop special interests that are not shared by peers, and this may exclude them from social interaction. For example, one 14-year-old girl with JNCL had a special interest in *Sleeping Beauty* and other fairy tales, but the classmates did not share this interest. Her participation in other class activities was reduced and the lack of shared interest made her generally more passive. This example depicts a problem, but also points to a solution: intervention that fine-tune or adapt the special interest may make it more inclusive. For example,

many fourteen-year olds have an interest in mythology. It is a short step from an interest in one fairy tale, to an interest in folk stories, legends and mythology from the world. This example illustrates that a small change may have cascade effects across a teenager's social and psychological development or maintenance.

Other students with JNCL develop special interests and advanced knowledge in areas that others in the class are interested in and which may in fact be relevant for the whole class. They can contribute to classroom activities provided the teachers implement the necessary measures and integrate the special topics of the students with JNCL in the work of the whole class. Music is such an interest that many children and young people share.

Within school, socially she does have a little peer group and at break time, she does have lunch break and morning break, you know, she'll look for someone to maybe go outside with her to get some fresh air and she has friends who are also interested in music and they tend to sit and chat about music. So she does have quite a good quality of social life in that respect.

The reduction in social participation may thus partly be a result of the students with JNCL not participating in the peers' common recreational and leisure time interests. Young people with JNCL may lose contact with the youth culture that exists outside the school and have no part in what their schoolmates are preoccupied with. This can easily become a negative spiral that may lead to

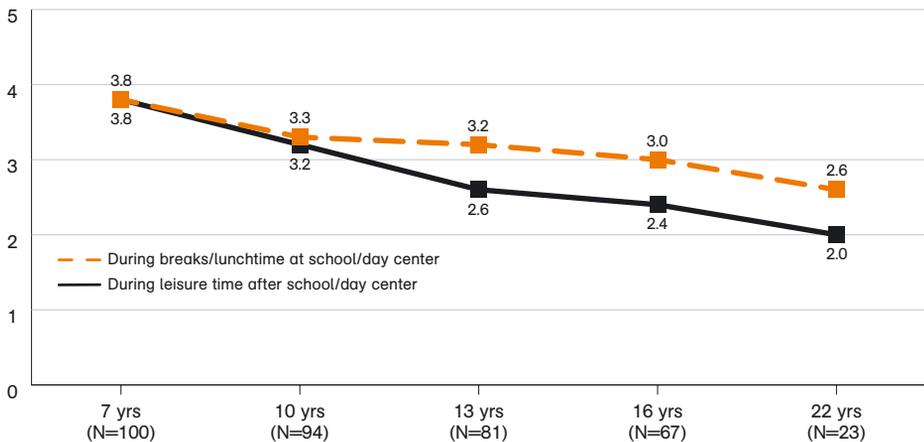


Figure 22.2 Interaction with peers of same age during breaks at school or lunch time at day center, and in leisure time outside school and day center

Scale: 1 = No interaction, 2 = Minimal interaction, 3 = Reasonable interaction, 4 = Pretty good interaction, 5 = Very good interaction

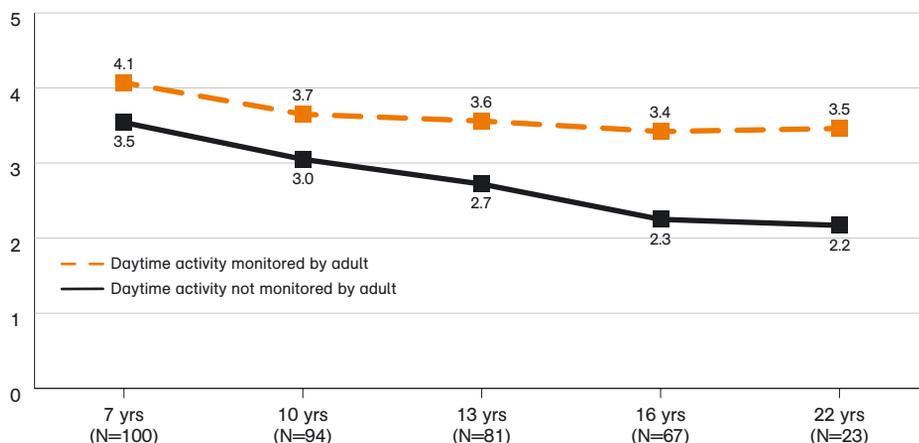


Figure 22.3 Social interaction with peers with and without adult monitoring across ages

Scale: 1 = No interaction with peers, 2 = Minimal interaction with peers, 3 = Reasonable interaction with peers, 4 = Pretty good interaction with peers, 5 = Very good interaction with peers

isolation of the student with JNCL both at school and during leisure time. Schools should therefore give priority to helping students with JNCL to participate in social activities and events. When teachers focus on themes and activities that may engage both the student with JNCL and the schoolmates, this can facilitate interaction between them. This requires that the teachers and other staff at school seek to identify possible shared interests.

The results of the present study (Appendix A) indicate that adult support and adaptation are crucial for social interaction between students with JNCL and their peers. The parents were asked to evaluate social interaction with peers with and without adult monitoring across ages on a scale from 1 (no interaction with peers) to 5 (very good interaction with peers). The importance of school as a social environment is documented by the fact that interaction with peers outside school and day center decreased faster than interaction with peers during breaks at school or the day center (see Figure 22.2).

Furthermore, interaction with peers was on average higher in activities monitored by adults and also the slope or the decline was smaller than in activities without adult monitoring (Figure 22.3)

The role of adults in monitoring social relationships was also mentioned by parents.

All social participation is managed by staff and other adults.

It is important to emphasize that reduced participation was not a result of a lack of motivation. Most of the participants with JNCL in the present project expressed a desire for social participation with peers.

And she would seek people out if she didn't know where they were. If it was particular people she wanted to chat to.

Her social interaction with peers is very limited. She does not approach peers in school and expresses a wish for being together with her boyfriend who attends the same school.

She has this befriender who comes and spends time with her. I think she is been shopping with her and she loves that. So she does arts and crafts with her and then mum takes her to see her friend and Tim so that is quite a good social thing. And she has tea with his family and things; she's quite a part of that as well. So yeah.

Several parents said that their child found the social loss the most difficult and emphasized that this situation should be remedied to the extent possible.

Adolescence is a particularly difficult time for young people with JNCL. The emerging cognitive, speech and motor declines become more severe, while social relations become more intimate and conversations more complex. In addition, adolescence represents new interaction patterns and romantic relations (Dunn, 2004).

Tommy lost his social life when he and his friends entered puberty.

She was able to keep her contacts to peers – two friends from elementary school – until she was 15 or 16 years old. She showed an extraordinary social competence and recognized the needs of others. When she was 18 or 19 it became very difficult and she frantically tried to keep in touch with people of her age. This didn't work out in the end.

The family is the social anchor for children and young people with JNCL, and with age the role of the family becomes even more important.

Ruth has no friendships outside the family.

Helmuth has no friends outside of his own Family. He has one roommate and they are good friends. The roommate has JNCL too.

In general, there is a reduction in peer interaction and social participation but it is still a varied picture that emerges.

She's got a nice group of friends that are very important in her life you know. She's got one boy and he teaches her drumming at lunchtime. She laughs a lot because school is a lot of fun for her. She's in touch with lots of the pupils and ex pupils on her phone at night phoning and emailing people so you know there's a lot of contact outside school and also with pupils that have left. It's quite nice that she is still in contact with them.

Including stories about romantic relationships.

She has a boyfriend who left school last year and the families keep in touch and he lives maybe about 30 miles away so I know that they go and spend weekends together, the families, so she gets to see her boyfriend and spends time with him.

These examples are important because they show that the reduction in peer interaction is not absolute, and some engage in romantic relations. Although peer relations in general were weakened, this was not always the case.

Social adaptation and intervention

The comments cited above suggest that it may matter what kind of support is provided to the young person with JNCL and that it may be possible to make adaptations and give support that contribute to maintaining social relations longer. The fact that it may be difficult to establish new social relations implies that trying to maintain some continuity in existing peer relations should be a main focus. This is also illustrated in the story about the Danish summer school below. When the parents were asked about the interaction with peers (Appendix A), there was a clear developmental pattern.

There were only few pupils in the class and the children and adults knew each other in a familiar environment. Interaction was good but when in a less familiar setting she was shy and liked to observe.

His social relations should be maintained with help from the staff, when he cannot do it himself.

In this example, the peers – with or without support – have found a way to compensate for the person's language decline and maintain interaction.

Social interaction is initiated by peers asking general questions around the person's favorite topic – high school musical or favorite sporting team. No initiation on the part of the person.

Similar strategies might be introduced to classmates by the teachers as declines become noticeable to both teachers and classmates.

Bullying also happens, maybe due to a lack of knowledge about the disease and its consequences, and it may be necessary to interfere in the peer environment.

The person's peers are generally accepting and tolerant however the first year of high school was very difficult. Some of the peers were very intolerant and would actively tease and try to cause anxiety in the person. This resulted in a new set of classmates for year eight which has been a huge improvement.

In school the breaks are important social events and school may make special arrangements to utilize the breaks as inclusive windows.

When Martha with JNCL began to lose her vision, the school established a system of break friends. Peers who wanted to spend some breaks with Martha put their name on a list. This became quite popular among the girls and they decided themselves that they wanted to be on the list. In the higher grades, boys in the class also asked to be on the list. Martha was very happy with this arrangement and asked every day who should be together with her in the breaks.

In addition, peer interactions may disclose issues that should be addressed in the education.

Martha's speech was not affected but she was not always aware of what was acceptable to talk about. The classmates were sometimes embarrassed when she talked about her own body and other private and intimate things. Observing this, the teacher initiated special sessions with Martha about advisable topics for conversations with break-friends.

Martha's story may also demonstrate the importance of peer continuity. It would probably have been much more difficult to establish a system of break friends if she had been new in the class and the classmates had not known her from the first grades.

The Danish summer school

As self-initiated and independent social participation gradually become difficult, the importance of organized activities increases. This part describes a Danish initiative to bring children and young people with JNCL together in shared activities and opportunities for social networking. Here is an invitation to our summer school experience!

One morning at the summer school: come on in!

Bacon and eggs are ready, together with coffee, juice and lots of Danish pastries. Of course, there is also oatmeal, hot and freshly cooked, standing steaming and ready to eat. The oatmeal is a special request from Ole. At home he eats oatmeal every morning, except for Sunday, when Ole, together with the family, eats bread from the baker. This is what makes Sundays special.

At all the tables everyone is eating. There are special plates where the edge is high and the food therefore stays on the plate even if the person is blind and has trouble with the fine motor skills. But first and foremost, this is a place with a great atmosphere; the room is filled with laughter, talk, lively debates about whether there will be an evening fire today or tomorrow, and each new entrant is showered with «good morning» greetings from all over the room.

During the meal, Peder stands up. Peder is Robert's assistant, or "supporter" as they are called in this assembly. And support is a good term; it means being the eyes, hands, body and memory for a child or a young person with JNCL. When Peder takes his place with a firm and safe hand on Robert's shoulder, this means that Peder and Robert are in charge of morning gymnastics. Or as Peder says: «Now, Robert wants to do morning gymnastics with you». Robert sits next to his chair with a big smile.

Anyone who can stand must get up, and then there will be lifting shoulders, shaking bodies, clapping, and all the hands are directed upwards as if trying to reach the ceiling, but nobody is that tall. A final highlight of the gymnastics comes when everyone is jumping on the spot making a good sound that is getting louder and louder as they stand there jumping. Phew! It is hard work at the morning gym!

Afterwards someone describes the weather and today's program. The task of recording "Today's sound" is allocated to one person and after dinner everybody tries to guess which sound was recorded during the day's activities. Then we are ready for today's. The first activity of the day is song and storytelling in the fireplace room. That is how a new day starts at the summer school.

JNCL Summer School

The NCL Team (see Chapters 9 and 24) organizes a one-week school event every summer for all children and adolescents in Denmark with JNCL. It is located at the Pindstrup Center in Djursland where there is ample space and a disability-friendly interior. Food and snacks are served throughout the day. There is a good atmosphere and helpful staff who will find scissors, paper and musical instruments from the stores when somebody has good ideas to be tested. The engagement and welcoming attitude of the staff at Pindstrup has a positive influence on the quality of the summer school. The rooms are distributed in the best possible way and special requests for food and drink are taken into account.

When the chef returned

For several years there was a chef at Pindstrup with a very special approach to the children and young people. He managed to create an atmosphere of peace and warmth. The chef could remember the children and young people from year to year, and it was clear that his presence became part of the summer school, activating both memories and energy. In 2016, for economic reasons, the chef had to leave and the food was delivered from the outside. This new arrangement was worrying, because it resulted in the loss of the former warm, supportive atmosphere for the children and young people.

However, the chef chose to visit the summer school in 2016, and his visit instantly created the good atmosphere and elicited many lovely memories. For the children and young people, and for supporters and consultants, it became an afternoon with a lot of smiles, and the chef reached all the children and young people with greetings and special remarks.

The summer school theme

The summer school is characterized by the pleasure of meeting new and old friends, learning and enjoying life. Every year, the summer school program is built around a special theme, such as "X-factor", "witches and trolls", "Nordic mythology", "Circus" and "Christmas". The basis for selecting a theme is that everyone in the group should be able to relate to it and contribute in accordance with their level of functioning. For example, everybody prepared for the great summer Christmas party, where everyone participated in singing and dancing with dresses and gifts.

Song and music

The summer school is characterized by song and music, which stimulate everyone's amusements and memories. A flexible Summer School Songbook has been prepared and has become something of a fond tradition. Old favorites are mixed with new songs, which are added as each new cohort of children adds songs to the collection. The songbook is much used throughout the summer school week and there is a special summer school song that is sung many times during the week. There is a great tradition of history reading and storytelling, and the program always includes afternoons with storytelling and songs where many express a wish for their favorite songs and stories from before and now, such as H. C Andersen's fairytales and stories from Nordic mythology, tales about Prop and Berta. All who can are singing. Most of the children and young people have favorite songs and they can sing or mime these even after their speech has disappeared.

Networking

A guiding principle of the summer school is the importance of children and young people being an active part of a social network. The summer school is a place where children can develop and sustain social connections; they get to know each other partly from prior summer schools, and also from regular activities sponsored by the Danish NCL Family Association. This network is often the only social network outside the immediate family circle that children and young people with JNCL maintain throughout their lives.

The support personell (supporters) of the children and young people with JNCL also form a network where experiences and best practices are shared. The summer school gives the support staff a lot of experiences in working with these children and adolescents, and for them the summer school represents a very useful «practice course».

Structure, predictability and the pedagogical foundation

Fundamental characteristics of the summer school are structure and predictability. There are sounds, pictures and many repetitions. The individual child or young person influences the program and how it is implemented. An important role of the support staff is to "translate" and interpret the wishes and tales of the children and young people. All children and young people have one or two supporters at the summer school and they are part of the long-term planning of the summer school as shown in the following excerpt of the invitation letter.

This letter to you as supporters is written because we from the NCL Team want to prepare the summer school in collaboration with you.

The role of the support staff in the summer school is, like in all the other network activities for children and young people with JNCL, to help to strengthen the fellowship among the participants including both the children and young people and the accompanying support staff. The summer school will in addition provide opportunities for learning a lot from each other.

One of the things we agreed to during the evaluation of last year's summer school was not to make the program too extensive and compressed. There must be room for inspiration and improvisation, creating good ideas on the spot and then pursue them, so the program for the summer school of this year is not so detailed. We want to allow room for the supporters and children to influence the content of the days – of course, based on the selected theme of the year. In that way, spontaneity and creativity will be the leading star of the forthcoming summer school and everyone are hereby invited to contribute to making the summer school amazing. The details of the individual days will be decided at our joint evening meetings.

We encourage you to prepare for summer school by talking to the children about the theme of the year (Christmas) and how they want it to be. How should the Christmas party be prepared, who would you like to be – Pyrus, Kandis or?

The Pindstrup Center has good facilities, so there will be ample opportunity to prepare this year's theme. The creative and practical preparations will take place in the workshop and on the first day, where we will delegate tasks and responsibilities, such as who will be responsible for each workshop.

You may think about everything from costumes to songs, tales, stories, and bring the things and props you might need. If you have a guitar or another instrument, please bring it – you do not need to be perfect on the instrument, we need every voice and tone! There is also a piano on the site. Bring songs that you sing in everyday life. We would like to learn new songs. And of course, there are also the songs in the Summer School Songbook!

Every evening there will be a gathering where we talk about the day and plan for the following days with support from the NCL Team.

We would like to talk with you about your role as a support person and how you can fill this role in the best way – that is, how you can contribute and strengthen the fellowship between all participants. As part of this, it is important that supporters who have experiences from previous summer schools share their knowledge and experiences with new supporters at the summer school.

Support staff at work

Like all other pedagogical work within the field of JNCL it is essential to consider the developmental decline, which sets its mark on the summer school program and the expectations of the support staff. The days have an overall structure and predictability, but the daily content is organized in a way that both welcomes and positively challenges the child and the young person with JNCL.

The Team and support staff work to create links between the new children and the group of slightly older children and adolescents. The supporters play an important role in this work, where traditions and practices are shared. Along the way, based on interests, there are reminiscences from former summer schools with songs, stories and repetitions.

The importance of the summer school for the participants with JNCL, for the support people, and for the NCL Team

The summer school offers meetings with old friends where memories are kept alive and old skills are refreshed. In the surveys and interviews with parents and staff (see Appendix A), tours and special activities were often mentioned as important events that their child with JNCL remembered particularly well. Two parents considered these events as one of the three most important activities or interests in the life of their child.

Working as a supporter for a child or young person with JNCL can be difficult but also rewarding with opportunities for learning. Children and young people with JNCL with a need for closeness and security make it necessary to work with professional values and reflections in collaboration with parents, other staff groups and the NCL Team (see Chapter 8). Supporters may experience many dilemmas in their daily work with children and young people with JNCL. The working conditions are not always consistent. It is important to establish support groups to encourage cooperation between members with different professional backgrounds. An important issue is how one "translates" and interprets the world to a child or a young person with dementia. Another issue is how to ensure that the selected goals and interventions reflect the true interests of the child and contribute to learning and good experiences.

The summer school emphasizes networking and learning in a community; that is, the summer school environment facilitates a transfer of important knowledge and best practice through the efforts of the supporters, who together create a good week for the children and young people with JNCL. The individual

supporters can act as role models for others in the network, ensuring that the children and young people with JNCL are in the center.

For the members of the NCL Team, the summer school week with children, young people and support staff provides a good opportunity to gain insight into the situation of each child and young person with JNCL. As a whole, the summer schools have an important function for the NCL Team, in relation to both the children and young people with JNCL and the continuous collaboration with the support staff who participates and receives guidance and supervision from the Team throughout the year.

Summer school in perspective

There are children and adolescents who have attended the summer school every year since they were diagnosed with JNCL. There are also children and young people whose parents have avoided the summer school and instead tried to bring the child into other social contexts, such as the summer school for children who are blind or have low vision.

There are many concerns for parents when deciding if their child should be a part of a community with other children with JNCL who have more pronounced declines and needs in later phases of the disease. There are no "correct" answers to these questions, families will differ in how and when they want their child to be introduced to the community of children and adolescents with JNCL.

At a recent parent meeting, the topics of the summer school and the children who did not attend the summer school were discussed. Several parents were curious, and also concerned. The NCL Team was asked to show the film that was recorded every year at the summer school to the parents who did not want to send their young children to the summer school. The parent of a 12-year-old said: *If you show the movie, everyone will understand how important it is for the child to attend the summer school as early as possible.*

Several of the video recordings from Summer Schools show young people without expressive speech "singing" when familiar songs were played. The young people were sitting and holding hands with friends from previous summer schools many years back. The young people had big smiles, despite serious losses of everyday skills and lack of control over body and facial expressions. Professionals have to learn to interpret what the children and the young persons are showing despite illness, declines and dementia. Participation and fellowship are important parts in the quality of life.

References

- Abdi, A. A., & Guo, S. (2008). Education and social development. In A. A. Abdi & S. Guo (Eds.), *Education and social development – Global issues and analyses* (pp. 1–12). Rotterdam, Netherlands: Sense Publishers.
- Bagwell, C. L., & Schmidt, M. E. (2011). The friendship quality of overtly and relationally victimized children. *Merrill-Palmer Quarterly*, *57*, 158–185.
- Barca, M. L., Engedal, K., Laks, J., & Selbæk, G. (2011). Quality of life among elderly patients with dementia in institutions. *Dementia and Geriatric Cognitive Disorders*, *31*, 435–442.
- Bukowski, W. M., & Adams, R. (2005). Peer relationships and psychopathology: Markers, moderators, mediators, mechanisms, and meanings. *Journal of Clinical Child and Adolescent Psychology*, *34*, 3–10.
- Bukowski, W. M., Motzoi, C., & Meyer, F. (2009). Friendship as process, function, and outcome. In K. H. Rubin, W. M. Bukowski & B. Laursen (Eds), *Handbook of peer interactions, relationships, and groups* (pp. 217–231). New York, NY: Guilford Press.
- Chen, X., French, D., & Schneider, B. H. (2006). Culture and peer relationships. In X. Chen, D. C. French & B. H. Schneider (Eds), *Peer relationships in a cultural context* (pp. 3–20). Cambridge: Cambridge University Press.
- Dunn, J. (2004). *Children's friendships: The beginnings of intimacy*. Oxford: Blackwell.
- Gilmore, L., & Cuskelly, M. (2014). Vulnerability to loneliness in people with intellectual disability: An explanatory model. *Journal of Policy and Practice in Intellectual Disabilities*, *11* (3), 192–199.
- Gold, D., Shaw, A., & Wolffe, K. (2010). The social lives of Canadian youths with visual impairments. *Journal of Visual Impairment and Blindness*, *104*, 431–443.
- Guralnick, M. J., Neville, B., Hammond, M. A., & Connor, R. T. (2007). The friendships of young children with developmental delays: A longitudinal analysis. *Journal of Applied Developmental Psychology*, *28*, 64–79.
- Hartup, W. W. (1999). Peer experience and its developmental significance. In M. Bennett (Ed.), *Developmental psychology: Achievements and prospects* (pp. 106–125). London, UK: Psychology Press.
- Hartup, W. W., & Stevens, N. (1997). Friendship and adaptation in the life course. *Psychological Bulletin*, *121*, 355–370.
- Laursen, B., & Pursell, G. (2009). Conflict in peer relationships. In K. H. Rubin, W. M. Bulowski & B. Laursen (Eds), *The handbook of peer interactions, relationship and group* (pp. 267–286). New York, NY: Guilford Press.
- Maccoby, E. E. (1990). Gender and relations: A developmental account. *American Psychologist*, *45*, 513–520.
- Mathur, R., & Berndt, T. J. (2006). Relations of friends' activities to friendship quality. *Journal of Early Adolescence*, *26*, 365–388.
- Meltzer, A., & Kramer, J. (2016). Siblinghood through disability studies perspectives: Diversifying discourse and knowledge about siblings with and without disabilities. *Disability and Society*, *31*, 17–32.
- Newsom, D., Mallow, J., & Watson, J. (2015) Loneliness in school age children: An integrative review of quantitative studies. *Psychology and Education – An Interdisciplinary Journal*, *50*, 32–41.

- Opie, J. L., & Southcott, J. (2018). Inclusion for a student with vision impairment: «They accept me, like, as in I am there, but they just won't talk to me». *The Qualitative Report*, 23, 1889–1904.
- Rossetti, Z., & Keenan, J. (2018). The nature of friendship between students with and without severe disabilities. *Remedial and Special Education*, 39, 195–210.
- Rubin, K. H., Bukowski, W., & Bowker, J. C. (2015). Children in peer groups. In R. M. Lerner, M. H. Bornstein & T. Leventhal (Eds.), *Handbook of child psychology and developmental science, Volume 4: Ecological settings and processes* (pp. 175–222). New York, NY: Wiley.
- Salminen, A. L., & Karhula, M. E. (2014). Young persons with visual impairment: Challenges of participation. *Scandinavian Journal of Occupational Therapy*, 21, 267–276.
- Schneider, B. H. (2016). *Childhood friendships and peer relations: Friends and enemies, Second edition*. Abingdon, UK: Routledge.
- Sterrett, K., Shire, S., & Kasari, C. (2017). Peer relationships among children with ASD: Interventions targeting social acceptance, friendships, and peer networks. *International Review of Research in Developmental Disabilities*, 52, 37–74.
- Wogn-Henriksen, K. (2012). «Du må ... skape deg et liv»: En kvalitativ studie om å oppleve og leve med demens basert på intervjuer med en gruppe personer med tidlig debuterende Alzheimers sykdom («You must ... make yourself a life»: A qualitative study on experiencing and living with dementia based on interviews with a group of persons who have early onset Alzheimer disease). Dissertation, Department of Psychology, Norwegian University of Science and Technology, Trondheim, Norway.

23

Transition to Adult Living

Svein Rokne and Bengt Elmerskog

Passivity, if life takes longer breaks and loses its fluency, if development and maintenance of skills of independence and interdependence are not given enough attention, and if participation in a socially inclusive environment is lacking, should be considered as threats for individuals with JNCL. These areas, often associated with the life flow, must be given special attention in situations of life transitions, and in particular to the transition to adult living.

Participation in various kinds of activities is mandatory for inclusion in the community and quality of life for all. However, there is a prominent risk that such goals are unobtainable for individuals with JNCL if their special needs are not considered and sufficiently met by appropriate measures. Involvement in *interactive participation* implies that individuals with JNCL can achieve personal goals when needs for personal assistance and physical adaptations are met by appropriate interventions. Interactive participation is closely related to personal *autonomy* (see Chapters 11 and 16). It supports personal engagement and applies to the personal rights of influencing one's own life, making one's own choices, having a meaningful work, having feelings of self-worth, being around people one likes, and having opportunities to enjoy experiences associated with learning and social and cultural living (Oakley & Marsden, 1984). Interactive participation is for instance used in theater plays where the audience is engaged in being an active part of scenarios (Skye, Wagenschutz, Steiger, & Kumagai, 2014). Interactive participation is an important strategy for empowering the individual, in particular in situations where the person's voice and interests should be articulated.

Interactive learning engages learners by challenging them to actively explore a material or situation. Through real-world applications, the individual makes meaningful connections between learning and its relevance for real-life situations. It reinvigorates the learning scene; one-way speeches or lectures are changed into

dialogues, discussions and collaboration. The learner and staff become partners in the journey of knowledge acquisition. It is found to be an effective way of learning, not only for people with learning disabilities, but also within higher education (Krusche, Seitz, Börstler, & Bruegge, 2017; Stanford University School of Medicine, 2018). Interactive learning can take many different forms. Individuals may strengthen their critical thinking and problem-solving skills using a holistic approach to learning in real-life situations. Interactive learning can thus have a place across the curriculum in a holistic manner. The close and timely interaction will promote immediate and specific feedback on comprehension (Stanford University School of Medicine, 2018). Interactive participation and learning are for individuals with JNCL closely related to interdependence where tasks are performed in collaboration with others (see Chapter 16).

Focus on participation in activities

A focus on participation in activities and participation-based learning has many advantages (see Chapter 11 and 16). Participation is an important basis for learning and development; children and adolescents learn from participating in activities – most knowledge and skills are in fact based on participation in real-life activities. For instance, children learn a lot through participation and exploration in preschool years. The learning is implicit and incidental, not based on predefined goals or curricula. This type of learning is valid for all humans, regardless of age and functioning, it is more about making participation possible.

The JNCL and education project (see Appendix A) has defined five areas of participation in activities to be considered when planning and implementing an optimal adult living: domestic activities, work activities, physical activities, leisure time activities, and vacation and excursion activities.

Participation in domestic activities is related to engagement in household activities, such as washing clothes, doing the dishes, cleaning, preparing food, and in domestic activities outside the home, for example shopping groceries and using the mobile telephone.

Participation in work activities is related to engagement in activities that give the young adult a social role in society or within a smaller group of peers. A working activity is associated with a clear and defined responsibility. This could include all activities undertaken in a vocational center or a sheltered workshop, such as metalwork, woodwork or pottery making where the products are available for sale either within or outside the vocational center or sheltered workshop. Participation in work activities usually take up a large part of the time spent outside the residence.

Participation in physical activities is related to engagement in activities that promote physical health and wellbeing. These are of special importance for individuals with JNCL and should take place on a daily basis. Physical activities may overlap with leisure activities, as when they reflect personal interests, but they should be defined as a separate category to ensure that they take place on a regular basis in order to sustain physical health and wellbeing. Physical activities can take place as organized activities (e.g., attending a sports club every Thursday) or as non-organized activities (a walk when the weather is fine).

Participation in leisure time activities is related to engagement in activities taking place outside work, school and domestic life, often during afternoons and weekends. Leisure time activities are strongly associated with personal interests and may be related to the person’s social needs (e.g., visiting friends or playing cards) and cultural needs (e.g., playing the piano, attending a football match or visiting the church). This category also includes certain self-entertainment needs such as playing a computer game, reading or listening to audio books, playing an

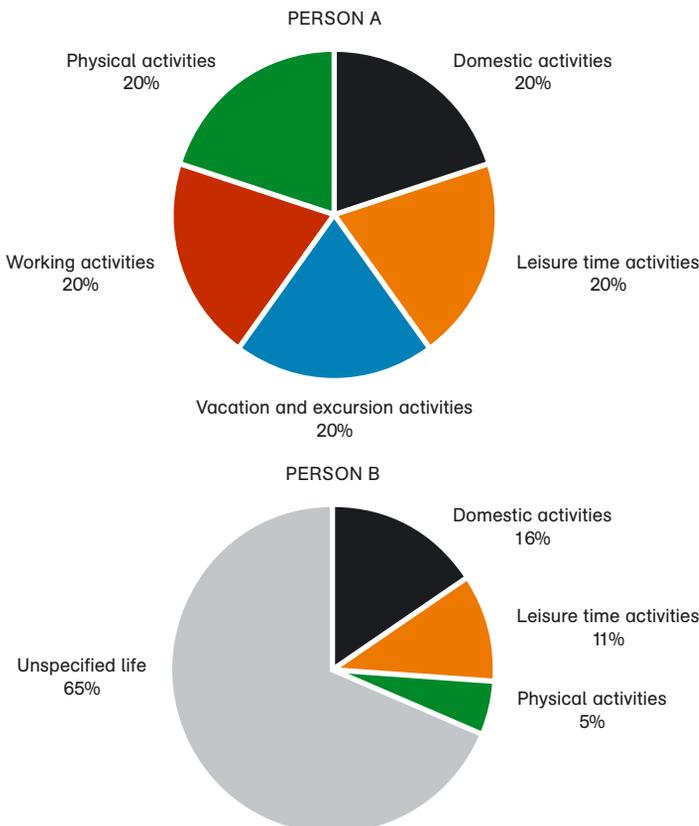


Figure 23.1 An overview of the life situation of two adults with JNCL

instrument or listening to music. Leisure time activities can be organized (e.g., participation in a book club) or unorganized (e.g., spontaneous activities such as listening to a CD to achieve relaxation).

Participation in vacation and excursion activities is related to engagement in activities, happenings or events beyond the daily routine. Vacation and excursion activities are strongly associated with personal interests that break the usual routine. A vacation activity can be a holiday trip to the mountains, a tour to a festival or visiting a museum in London. Vacation and excursion activities are considered as highlights taking place on an infrequent basis.

It is assumed that a reasonable balance between these activity areas will promote life quality and participation in the community. Participation in the different spheres of activities may function as an index of quality of life.

Figure 23.1 shows different life situations for two young adults with JNCL. Person A has a well-balanced life situation, while Person B has a limited life situation. The size of the sector named "unspecified life" seems worrying for Person B. A focus on participation in the different spheres of activities constitutes a fundamental basis to achieve personal goals associated with interactive participation and learning, in particular at the time of transition to adult living.

Participation when interdependence is required

Independence will be reduced as individuals with JNCL get older (see Chapters 3–7, 16). There is always a risk that participation will be threatened when abilities and functions decline. Gray and Hollingsworth (1999) developed a model for describing the relations between participation in activities and performance in activities. Table 23.1 illustrates four scenarios for the relations between ability, performance and participation in particular activities or tasks for one individual. *Scenario 1* is a rather normal situation where participation and independence are correlated. *Scenario 2* is a situation that may have been caused by poor planning in connection with the transition to adult living for a person with JNCL. For instance, the person was a devoted and skilled person in using the computer the last year in school. However, the person became excluded from this activity in the new residential home due to a lack of equipment or staff competence related to using the specialized equipment. *Scenario 3* may be positive for an individual with JNCL. The person is not able to do much but is provided with enough support to participate through interdependence. *Scenario 4* might be a situation where low performance automatically led to non-participation caused by contextual factors, such as the lack of support and help.

Participation in activities may over time become limited or take so much effort that the person with JNCL will not find it worthwhile to initiate participation in

Table 23.1 Four scenarios related to activity and participation (after Gray and Hollingsworth, 1999)

	Ability to perform an activity	Inability to perform an activity
Participates	<p>1</p> <p>The person has the capacity to do things and does participate</p>	<p>3</p> <p>The person cannot do things but participates</p>
Does not participate	<p>2</p> <p>The person has the capacity to do things but does not participate</p>	<p>4</p> <p>The person has no capacity to do things and does not participate</p>

some activities. The risk is however that non-performance automatically leads to non-participation caused by lack of support or other contextual factors. Scenario 1 and 3 in Table 23.1 can consequently be considered as positive goals for young adults with JNCL (see also Chapter 11).

Persons with JNCL are affected by declines that may threaten participation. The anticipation of these declines requires that the staff is introduced to and taught about the Team Model (see figure 23.2) which may help them to achieve continued participation when abilities decline in persons with JNCL. It is the way that the activity is organized that governs the situation. It is necessary to acknowledge that persons with JNCL at all times may participate according to their abilities, achieve personal goals and experience pride in spite of progressing declines in performance. The person with JNCL and a support person should work

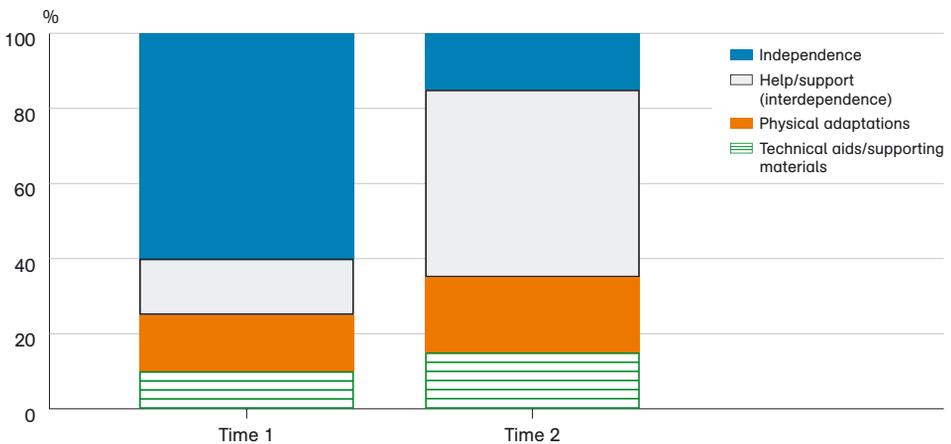


Figure 23.2 The Team Model or The Interdependence Model describing 100 percent participation at two times

together as a team, although the relative contribution from the person with JNCL and the supporter will change over time (see Figure 23.3).

The base line at Time 1 in Figure 23.2 shows that the child with JNCL performs 60 percent of the task while the supporter contributes 15 percent. Ten percent of the task is supported by technical aids and 15 percent by physical adaptations of the environment. At Time 2 the person is contributing only 15 percent of the task. However, the level of participation, perceived by the individual with JNCL, is 100 percent at both times. The participation level at Time 2 is based on more support, more physical adaptations and additional supporting materials compared with Time 1. (The two scenarios are illustrated in Figure 23.3.) It is thus important that the people who are assigning resources understand the necessity of establishing the Team Model when working with individuals with JNCL. The access to individual support or other adaptations will sooner or later constitute the difference between participation and non-participation for persons in this group. The Team Model can be applied to any situation, for instance at home or at work, as the parent story below illustrates.

Roger is genetically predisposed in more than one way; from his father he has inherited an interest in fly fishing and has found profound pleasure in catching representatives of the aquatic domain on flies tied by himself. He learned the basic techniques of fly tying quite early, while he still

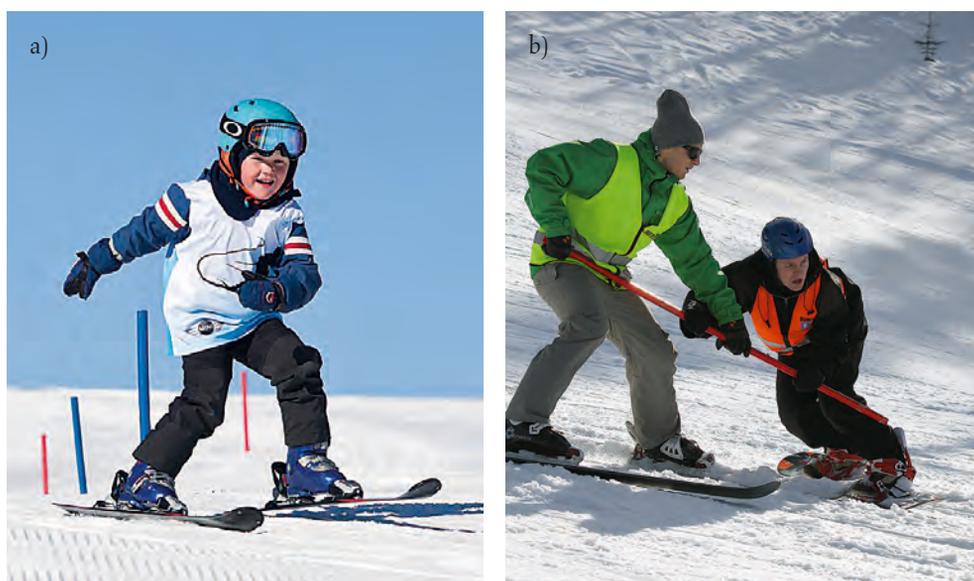


Figure 23.3 Participation through a) independence and b) interdependence

had some residual sight. When seeing became gradually more difficult, he appointed his father to the post of assistant fly tier, and the flies were produced through a joint effort.

Fly tying is a process requiring fine motor skills and the ability to consider and make decisions related to type and size of hook, tying pattern, choice of color scheme, hackle type, ribbing, wing type and more. These elements were discussed before starting the tying process at the tying vice. Salmon flies have always been central to Roger's fly tying, particularly since he had been able to try out the results of his efforts in the salmon rivers during the summer, often successfully!

When Roger was 20 years old, the idea to take the fly tying one step further materialized. Why not use the salmon flies as a basis for production of earrings? Roger's father was appointed as Roger's business manager and 5 percent of sales were considered a fair remuneration for this work (determined after tough negotiations). This resulted in a planning phase, where "Team Roger" had to consider and decide upon several factors related to production, buying, marketing, sales work, logistics, accounting, invoicing and managing receivables. A company was established: Roger's Products.

The process from hatching the idea to making the first sale (and collecting the money) was incredibly exciting and educational for Roger and his father. Standard patterns needed to be developed, fly-tying components had to be purchased (involving several visits to various pro shops for selecting the raw material), and solutions for clasping the product to the ear (not a trivial point) needed to be developed. In addition, a marketing plan was conceived, and Team Roger had a discussion whether to base the business on a one-tier or two-tier sales model. Also, a price strategy needed to be established, and then the actual marketing and sales work started.

An invoice is produced for each delivery, and Roger's father ensures that these are being paid and that the money thus accruing is deposited into Roger's account. Rest assured that Roger follows this part closely!

During the two to three years of production since the inception of the business, Roger's abilities have been somewhat reduced, meaning that more of the physical labor is carried out by the second participant of the Team. However, Roger owns the business, the production process and the sales efforts, and participates all the way. There has been no diminishing of pride each time Roger's Products produces and sells a new set of earrings to a satisfied customer.

From adolescence to adulthood

Reaching adulthood is an important milestone in anybody's life, also in the life of a person afflicted with JNCL. For individuals with JNCL, the transition period between adolescence and adulthood is of particular importance. The quality of their adult life will depend on several factors discussed in other chapters of this book and on how the transition period is planned and carried out.

What does it mean to be an adult? And more specifically, what does it mean to be an adult with JNCL? Most encyclopedias define an adult as a fully grown and developed person, mature and sensible – not childish. In the legal sense, an adult is a person who has attained the age of majority (or maturity) and is therefore regarded as independent, self-sufficient and responsible.

The first part of this definition is problematic as far as persons with JNCL are concerned. The progressive nature of dementia means that the definition is only partly applicable. In the legal sense the definition applies as long as the person has not been declared legally incompetent. In the present context, none of the definitions are of any concern. The goal is to examine the process of transition and recommend strategies which may contribute to the person's quality of life as an adult, which here means a person with JNCL who has finished secondary school, entered a place of work or is engaged in other suitable occupation, and who has moved out of the family home.

All the interventions, education, and training during the school years were intended to contribute significantly to learning and quality of life during these years, as well as to prepare the person with JNCL and adapt the environment to future adult living. A transition to adult life often means that the person with JNCL moves to a new environment, an apartment or as a resident in a sheltered house and community of peers. The person should in addition participate in some meaningful daytime work-related occupation, which is located outside the residence. These are important aspects of adult living.

Several considerations must be made before, during and after the transition period. This chapter is based on findings from the present study (Appendix A), including some recommendations to achieve *best practice* in times of transitions to adult living. Core factors behind successful transitions are a functional responsibility group (see Chapter 11), service providers' competence and attitudes, and available resources. There are vital differences between countries in terms of culture, how society is organized or how services for persons with disabilities are provided. However, the transition should always be person-centered, independent on the overall system or cultural attitudes. Individuals with JNCL share the disease but in all other aspects they differ as much as other people.

Findings from the JNCL project

The present study did not find any research or documentation related to adult living among people with JNCL that go beyond the obvious medical needs. Quality of life encompasses areas beyond medical health, and includes needs in relation to social life, life content, learning and maintaining skills and development. The findings of the present study (Appendix A) give a better understanding of the transition period from school to adult living and the factors that may contribute positively or negatively to the adult living for persons with JNCL beyond their medical needs.

There was a consensus among the parents that local competence had been lacking, not only in relation to JNCL or early onset of dementia, but also to meeting the challenges related to teaching a person who is blind. Particularly problematic was the comprehensive change of staff and external counseling when the person reached legal age, which in most countries is 18 years.

A majority of the parents of persons who had reached the age of 22 were reasonably satisfied with their collaboration with the local community, but fewer with local education counselors and, perhaps surprisingly, educational counselors in resource centers. These responses can indicate a lack of sufficient competence or of access to competent services.

Interest in physical activities was low from 17 years onwards, although parents seemed to agree that this is an important area for their child (see Chapters 7 and 15). The results show a significant reduction in physical activities as the person gets older. One of the reasons may be that while physical activity is emphasized as an important part of the school curriculum, it becomes more difficult to implement physical activities in the family home or in residences for adults with disabilities. The lack of physical activity can also be due to a lack of knowledge about how to achieve a healthy lifestyle when the person is blind and has reduced motor functioning.

When evaluating the various school subjects and their impact on life as an adult, the parents considered music, music therapy, physical education and physiotherapy as most important. Literacy, handicraft, cooking, kitchen work, social training, occupational therapy and social gathering were considered as having a positive impact but were given somewhat lower scores. Mathematics, social studies, nature studies and foreign languages were given low impact scores by the parents. A large majority (90%) found that augmentative and alternative communication training had little or no impact on their child's life after the conclusion of school but only a small number of persons had received this form of training (see Chapter 13).

Encouragement of special interests or hobbies and the integration of these into adult living had been lacking according to parents. Early establishment of skills and a continuous maintenance of these skills seem to give better chances for these skills to be maintained into adulthood. Examples from the present study are playing an instrument, horseback riding, swimming, music and dancing. Computer skills and literacy are also mentioned as long-lasting skills – reading and writing probably had a positive influence on the preservation of cognitive skills. Many parental comments suggested that having a suitable daytime occupation after school helped to maintain skills for a longer period.

Asking staff the same questions yielded the same results as far as music, physical education and physiotherapy are concerned, while their impact scores for reading and writing were very low – they found that these subjects had little impact on life after school for the participants with JNCL. Comments point to a feeling of defeat and, perhaps, little understanding of the necessity of continuous stimulation to maintain skills. A descending curve of abilities should not be used as a reason to terminate initiatives to measures that may maintain skills in young adults with JNCL.

A large majority of the parents (86%) agreed with the statement that the school had done reasonably well in preparing the individual with JNCL for adult living. Sixty-five percent of the parents agreed that the school did well in transferring important information to the person's new environments (home and day center), and a majority (68%) also agreed that the home or day center had been reasonably well prepared. A majority (68%) agreed that the collaboration between parents, the school and staff in day centers or homes had been successful, whereas 59 percent were satisfied with the role external counselors had played in this process. However, some parents expressed dissatisfactions with the transition period as shown in the following quotation:

It is important to emphasize the transition to adult living for the school. For us nearly two years have passed since end of school and not much is in place.

Seventeen of the fifty responding parents reported that they were offered a sheltered housing or similar after the conclusion of school and 39 of the persons with JNCL had been offered to join a day center or similar. It seems that housing remains a difficult issue.

Although their child's transition to adulthood and living as an adult had been, and remained, quite challenging, several parents reported satisfaction with the process as well as with the period following the transition. Some parents expressed satisfactions with highly motivated service providers, who did their work with the right kind of attitude and often beyond their formal responsibilities.

The preparatory phase

A long planning period may be required to find a home and an occupation for a person with JNCL, depending on several factors. The transition period is the time from initiating the transition project (at least two years before school ends) to the actual milestone: Starting work-related activities and possibly moving to a new home. The strategies, activities and tasks in the transition period should be described in detail in the long-term Habilitation Plan developed by a Responsibility Group (see Chapter 11). The plan should list tasks, responsibilities and success criteria with associated dates. The process should be monitored through frequent evaluations, at least once every third month during the transition period. The local government may assist in this and the municipality usually has a planning period

Table 23.2 Topics that may be included in the Habilitation Plan

Topics, needs and functionalities for the future	
Network	Family, friends, care givers
Daytime activity	Work, activity center, other
Leisure	Holidays, daily activities, sports, hobbies, areas of special interest
Preparatory training	Training at current school to prepare the individual for adult life in the forthcoming home, day center or work? How? By whom?
Living preferences	Physical layout, own room, adaptations needed, the neighborhood
Aids	Computer equipment, wheelchair, other aids.
Communication and language	How does the person communicate? Tools to aid communication (manual signs, communication aid, computer, other)?
Behavior, social life	Resources, strengths, vulnerabilities, wellbeing, play, mood, frustration, aggression, withdrawal, sleep pattern, depression, anxiety
Nutrition	Dietary requirements, meals, appetite, food and fluid intake
Motor skills, disabilities	Mobility, gross and fine motor functions, technical aids
Support	Support needed (individual or shared)
Health	Diagnosis, medication, epilepsy, teeth condition, bowel function, sight, hearing, allergic reactions, contingency plans, etc.
Economy	Legal rights, social security, other
Aid measures	Educational and other services provisions, scope of services rendered, other useful information
Staff competence	Competence building. How, when, and by whom?

of several years. The findings of the present study emphasize the importance of starting this work early.

In addition to, and as an integral part of the Habilitation Plan, it will be of great help to have a written narrative of the personal history of the individual with JNCL. This written narrative – «The book of (the person's name)» – is regularly updated. This is a valuable tool for guiding service providers and staff to a shared frame of reference. The Responsibility Group should consist of individuals who are able and willing to carry out the plan. One member of the group should act as the group's coordinator and have the additional responsibility of organizing meetings, setting the agenda of the meetings, and monitoring the work progress in general. This also includes measuring results compared to the goals of the plan and evaluating the overall strategy. The Habilitation Plan includes functions and needs in all areas of life (Table 23.2).

The future home

A good beginning would be to design the "perfect" future home for the person with JNCL; considerations should include the physical location and the quality and quantity of service provisions needed in the new home. Designing a new home two or more years into the future is challenging. The person with JNCL may at the onset of the transition period be functioning quite well and with some degree of independence when performing daily tasks. However, there is a need to consider needs that are likely to appear years ahead and design the optimal home with those in mind. The local government may assist the parents (and other family members) in designing and finding the new home.

There are several possible strategies for identifying the new home. One strategy is to apply to an existing house in which a group of young adults with disabilities have their own facilities surrounding common areas such as community kitchen and drawing room(s). The new home should be adapted to the needs of persons with visual and physical impairments. For instance, it should not have physical barriers such as steps or dangerous indoor and outdoor environments. Young adults with JNCL may also be sensitive to neighbors with challenging behaviors and environments with high noise levels. The rationale for moving into a community is usually to create opportunities for social interaction with peers. The current study shows however that persons with JNCL over time tend to spend more time with caregivers and staff and less in social interaction with peers. Loss of friends is an unfortunate fact of life for most individuals with JNCL (see Chapter 22), and planning should take this into consideration.

Another strategy is to move into an apartment which can also accommodate one or more staff as the need arises. However, finding a suitable apartment can

be difficult and costly. A third strategy may be to rebuild the family's house to accommodate a separate home for the individual with JNCL. Proximity to parents, siblings and other family members may have benefits as well as disadvantages, for the person as well as for the family. Finally, in some countries there are special institutions that cater to the needs of individuals with JNCL, such as Heather House in the U. K. and the Bartimeus Foundation in the Netherlands.

It is not always possible to find a home that matches the needs of the individual. The result may be that the person with JNCL continues living with the parents for a shorter or longer period after finishing school. However, if a suitable home is found, it must be furnished with care and on the basis of the person's anticipated physical and mental condition at the time of moving.

Moving out of the family home should be a gradual process, where the person moves in steps, beginning with shorter day visits accompanied by parents, which are gradually expanded into overnight visits. Comments from parents and staff in the present study emphasize the importance of preparing the young adult with JNCL for the transition by talking about it, and when possible – organizing many visits to the future home. The young adult will become familiar with the layout of the apartment or rooms and may become acquainted with the other residents (if any) and the staff before moving there. Such an approach reflects that moving is a process and may make the move more interesting and feasible for the person with JNCL.

In some European countries, young adults with JNCL receive a pension for persons with disabilities from age 18 years and onwards. Together with other forms of economic support from the government (basic and extended economic compensatory benefit, living cost subsidy), the young adults will usually be able to fend for themselves from an economic point of view. In some countries, purchase of living facilities, if necessary and desired, may be subsidized through low-interest loans and grants from the government.

The future occupation

Finding a suitable occupation at a sheltered workshop or day center after leaving school is of extreme importance. The last years at school should include vocational and work-related activities to facilitate the transition to adult life. One or two days should be used to train the person in occupational activities similar to those that may be provided after the young person leaves school. Consequently, the identification of an appropriate occupation should also be discussed early in the transition process. In the present study (Appendix A), some parents reported that the student with JNCL had been offered opportunities to work one or two days per week at their future adapted workplace as a part of their Individual Educational

Plan (IEP, see Chapter 11). According to the reports, such educational planning measures made the transition much easier.

Although blindness and cognitive and physical declines may limit the availability of job-related activities, no efforts should be spared in securing a suitable occupation. Comments in the present survey and interviews indicate that getting up in the morning and going to a different location for work-related activities seemed to have significant therapeutic effects, including heightened self-esteem, particularly if a small remuneration was attached to the work. In addition, the understanding of The Team or Interdependence Model (see Figure 23.2) will simplify the process of finding a suitable occupation. Team members should consider what the person may be able to do or produce when he or she is collaborating with a support person. The concept of supported employment relies on the concept of interdependence.

The challenge, of course, is finding a meaningful occupation for the person. Ideally, the person has skills and preferences that can function as a basis for working activities. It will be useful to arrange meetings with representatives from the local government and work places early in the process. Most municipalities have sheltered work places catering to the needs of young adults with disabilities, and many activities in such places can be tailored to fit the needs of a person with JNCL. Meetings with representatives from these work places may create an understanding of the needs, that is, the anticipated progressive decline in abilities and how a work place can cater for such needs with an approach based on interdependence (see Chapter 16).

Staff and service provision

When planning the transition to adult life, it is necessary to determine what resources are necessary to adequately cover the person's needs once he or she moves to a separate home and starts the working life. The course of the disease, in particular the fact that epilepsy may become a severe problem, calls for proximity to staff around the clock. As soon as adequate housing has been found, an application must be presented to the local government for necessary staff at the living facilities. How this is organized in practical terms may vary considerably, not only from country to country, but also within countries and communities.

Service provision for young adults who need continuous staff attendance is covered by several laws. However, because of the broad leeway often given to how the laws are interpreted, the process will not always lead to good solutions. Often resolving the problems resulting from deficits in the process entails hard work for the parents with or without assistance from resource centers or parent organizations. For example, although the legal process typically recognizes the

need for continuous medical care, it often fails to recognize the resources needed to maintain an optimal quality of life. In view of the progressive nature of JNCL, the human resources required for maintaining the goals of interdependence and autonomy (see Chapter 11 and 16) will gradually increase, and planning must take this into account.

Local governments often have preferences as to how services should be organized. Most local governments in Norway recognize User-Managed Personal Assistance as a way of organizing services. Under this scheme, the user, in this case an individual with JNCL, is *de facto* the employer and must hire personnel to provide services within an economic frame decided by the local government. In theory, this gives some degree of quality control, but in general, user-managed personal assistance represents extensive administrative work for the parents. Most often, however, services are provided by the local government. The advantage is a reduction in the administrative burden of the parents, but the disadvantage is the potential risk for compromises in the quality of the services provided to the person with JNCL.

If the parents wish to continue being the guardians for the individual with JNCL, it is a good idea to issue an authorization from the person with JNCL to one or both parents, authorizing them to act on the person's behalf in all matters, economic and otherwise. The authorization signature (made with or without help) should be confirmed by two witnesses. If the parents do not wish to have this role after the person reaches legal age (18 years in most countries), they should apply for a guardian to safeguard the person's rights.

Educating staff

When an appropriate home for the person has been found and the time to move is getting closer, the future staff should be educated and informed to avoid unnecessary problems in the moving process. An appropriate working culture should be encouraged from the very beginning. Parents and staff in the present study (Appendix A) had found the use of person-centered work-shops to be a good method to educate staff who will be in direct interaction with the person with JNCL. The workshops should focus on possibilities instead of limitations. Getting to know the person rather than the disease should be the main focus of the workshops. Education of staff is an ongoing process, for instance built on the stepwise method presented below. The first work-shop may be based on *The book of (name of the person)* (see Chapter 11), preferably presented by the parents, aiming to make the staff comfortable with their roles in supporting and communicating with the person. An early open dialogue between parents and staff will contribute to building a foundation for further collaboration and competence building.

Life as an adult

The quality of life as an adult depends on many factors. Early established skills and interests can often be maintained in a longer perspective and thus contribute to a richer life for the young adult. The implementation and quality of the preparatory phase is another important factor, including the establishment and maintenance of good relations between the family, staff and the person with JNCL.

Teamwork

An optimal adult living for a person with JNCL will be based on the coadjutant equal partners: parents, staff, representatives from the local government and counselors with knowledge about JNCL. The team must find a good and effective way of working together, based on mutual trust and efficient communication. The meetings can be held as workshops where all participants are given the possibility to learn, share knowledge and promote ideas. Minutes should always be kept from these meetings and agreed actions should be clearly spelled out.

The team may have chosen a place to live with a (small) number of peers occupying their own facilities around a common kitchen and a drawing room, and with staff in accordance with the person's needs and as determined by the local government. The team should have agreed on the important principles or framework for the work. The principles agreed by the team should emphasize maintenance through daily or weekly repetitions of acquired skills and knowledge. Staff turnover should be minimized, and an effort to maintain a core staff group responsible for the person should be made so that the person's daily life routines can remain as stable and predictable as possible. High staff turnover is a reality in the field, and a plan for continuous training should be in place for new staff. Likewise, a system should be in place for providing substitute staff members with the required basic knowledge when needed. Daily physical and mental exercises and challenges should be managed in accordance with the goals defined by the team. The daily routines should be followed by all staff members, even in times of worry and pronounced health problems.

A method for supporting adult living

A method for establishing the best possible adult living for persons with JNCL has emerged from the quantitative and qualitative results of the present study. The method does not focus on the medical needs but on the needs associated with content of life. The establishment and maintenance of rich and varied life content are essential factors that will contribute to the quality of life and general

health of persons with JNCL. There is an emphasis on daily cognitive and physical activity and learning and maintenance of skills from early morning to bedtime. The method includes several tools.

Tool 1 – The book about (name of the person)

A continuously updated written narrative of the person's history is an integral part of the present method. The content of *The book about (name of the person)* includes historical and present facts about the person with JNCL and an overview of important topics, areas of interest and preferences. Having access to this book makes it easier for others, specifically supporters, to understand the state of the person at any given time and support an active life style. *The Book of (name of the person)* is a central document for service providers and mandatory reading for all staff working with the individual with JNCL. Provision of time for supporters to read the book might be one of the overlooked planning resources described earlier. The book is a valuable tool for new personnel and regular reading may serve to refresh the memory of those who already know the person. The book is introduced as part of the preparatory phase (see above) and remains a part of the Habilitation Plan. The aim is that the book should contribute to promoting an understanding of the person with JNCL as a unique individual with a personality and history despite current and future declines (see Table 23.3). It should help establish a shared frame of reference between the person and the staff, and between staff members, thereby facilitating communication. The book should not only make it easier for staff to maintain focus and engagement, but also provide them with a way to enrich their own work days by enabling them to experience success doing their job. It is easy to overlook the daily challenges staff may face, not just from doing their duties, but also from external pressure and demands from family and institutions.

Tool 2 – The Annual Wheel

Every year there are important events and activities taking place at regular or irregular times, such as vacations or birthdays. *The Annual Wheel* is used to identify such events and activities. Participation in vacation and excursion activities is related to engagement in activities, happenings or events beyond the daily routines. Vacation and excursion activities are strongly associated with personal interests that break the usual humdrum routine. A vacation can be a holiday tour to the mountains, an excursion can be a tour to a festival or museum in London. These vacations and excursions are typically highlights in the ordinary time flow, taking place infrequently. Figure 23.4 shows an Annual Wheel for a young adult with JNCL. *The Book of (name of the person)* and the parents should be consulted when constructing an Annual Wheel.

Table 23.3 Possible contents of The book about (name of the person)

Topics, needs and functionalities for the future	
1 – Why this book?	A brief introduction to (name) and his or her special situation and what his or her condition means in terms of necessary requirements and desired skills in daily interaction with him or her. A list of the most important elements contributing to the person's quality of life can be included here. Why is it necessary to have this book, what does it include, who is it for and how should it be used?
2 – Who is (name)?	A brief life history
3 – (Name's) routines	Daily and weekly routines
4 – To lose one's sight	What are the consequences of blindness? How does (name) cope with this situation and how can service providers help. Writing/reading with Braille or Moon. Computer tools, daily living aids, mobility issues.
5 – Medical conditions	Emphasis on the here and now and on symptoms rather than the diagnosis. Medication and how it is administered. Reference to contingency plans.
6 – Communication	How does (name) communicate? Any differences between expressive communication and comprehension?
7 – What (name) is concerned about	Skills and learning, hobbies
8 – Interests	All areas of interest and how best to stimulate the person through active use of these. Most individuals with JNCL are interested in music, and early learning for example of piano playing, drums or other instruments can be maintained for a very long time.
9 – Important persons in (name's) life	Family members, important friends, others
10 – Important places for (name)	All important places and why they are important. Home, work place, places of important activities, travel destinations. Can provide common ground for interaction and communication.
11 – Important activities	Activities contributing to the person's quality of life, summer and winter activities, programmed activities as well as activities of a more ad hoc nature.
12 – Aids	For example wheelchair, tandem bicycle, winter and summer sports equipment, kitchen aids, the white cane, telephone, MP3 players, and camera.
13 – Calendar	All important regular and irregular events, such as birthdays, vacation plans, or work schedule
14 – Food and drink	What the person likes to eat and drink for breakfast, lunch or dinner, and special dietary needs
15 – Personal hygiene and need for assistance	What the person can do with or without assistance. Routines for morning and evening; special needs in the shower, special toilet requirement, clothing
16 – List of names	All important names, telephone numbers, mail addresses, who does what
Addenda	For example, general information about JNCL, information about epilepsy and medication, or medical contingency plans

The Annual Wheel Important events throughout the year not appearing on regular basis (birthdays, vacation etc.)			
Name of Person: Erik Jenssen		Year: 2018	
Specification of activity or event:		From:	To:
January	Dad's birthday	10/1/18	10/1/18
	Weekend with Mum and Dad	19/1/18	21/1/18
	Winter camp - Hurdal Center	22/1/18	27/1/18
February			
	Weekend with Mum and Dad	23/2/18	25/2/18
	Participation in Roeros Festival		
March			
	Sister Ida's birthday	21/3/18	21/3/18
	Participation in Melhus masquerade ball	23/3/18	25/3/18
	Weekend with Mum and Dad	24/8/18	26/8/18
April			
	Easter vacation (not specified yet)	9/4/18	15/4/18
	Weekend with Mum and Dad	27/4/18	29/4/18
<i>and so on.....</i>			

Figure 23.4 An Annual Wheel

Tool 3 – The Activity Bank

The activity bank contains activities related to the five areas of participation in an optimal adult life described earlier: (1) domestic activities, (2) work activities, (3) physical activities (4) leisure time activities, and (5) vacation and excursion activities. The activity bank is supposed to be used for activities that may change from day to day but which are of special importance to the person with JNCL. The aim is that the young adult with JNCL should be engaged in all the activities in the activity bank on a regular or irregular basis. Figure 23.5 shows the activity bank for a young adult with JNCL.

The *Book of (name of the person)* and the parents should be consulted when constructing the person’s activity bank. It is a vital source of information for staff both in the residence and at the working place, for instance for promoting sufficient physical activities to achieve the best possible physical fitness or for promoting or maintaining domestic and cultural skills.

Tool 4 – The Week Plan

The *Week Plan* is a week schedule showing the structure of the routine activities taking place every day (Figure 23.6). Some of the activities in the Week Plan are however open or not predefined and should be filled with activities from the *Activity Bank*. These activities can change from day to day according to the person’s wishes or needs, for instance in accordance with the goal of staying physically and cognitively fit or to maintain skills and interests in music.

Participation and Activity Bank				
Name of Person: Erik Jensen		Year: 2018		
A. Leisure time activities	B. Physical activities	C. Vacation and excursion activities	D. Domestic activities	E. Work related activities
1 Play Cards	1 Walking tour	1 Concerts	1 Go shopping	1 Sort screws and nuts
2 Play Board Games	2 Physiotherapy program	2 Café	2 Set the table	2 Polish cups and plates
3 Play Piano	3 Weight-lifting program	3 Restaurant	3 Cook simple food	3 Knitting machine
4 PC games	4 Visiting the gym	4 Fishing	4 Cut the vegetables	4 Make fire briquets
5 Listen to Audio Book	5 Swimming	5 Visit Parents	5 Empty the garbage bin	5 Empty paper bins
6 Listen to Music	6 Aerobic program	6 Visit sister Ida	6 Clear the bedroom	6 Fill firewood sacks
7 Make Music	7 Tandem cycling	7 Picnic	7 Do the dishes	7 Transport firewood to store
8 Singing	8 Mountaineering wall	8 Visit Grandfather	8 Make the box lunch	8 Sell firewood at market
9 Play the drums	9 Ski sledge	9 Winter camps	9 Bring the post	9 Prepare lunch to fellows
10 Play the harmonica	10 Wheelchair exercises	10 Summer camps	10 Make shopping plans	10 Make candlesticks
11 Call family and friends	11 Relaxation exercises	11 NCL peer meetings	11 Make coffee/tea to others	11 Parcel candlesticks
12 Check/write E-mails	12 Articulation exercises	12 Visit children’s museum	12 Fetch firewood	12 Fill paper - copying machine
13 Socializing with neighbors	13 Stretching exercises in bed	13 Vacation trip to Spain	13 Dry the dishes	13 Label the products for sale
<i>And so on.....</i>				

Figure 23.5 An Activity Bank

Tool 5 – Registration of Participation

In the present study (Appendix A), many parents commented that they were barely aware of what was happening to the person with JNCL in the new home or at work. Some parents expressed frustration that the level of participation was lower than the expectations, as in the following quotation:

They said they were going to visit the indoor swimming pool on a weekly basis, but this has not happened so far.

Making daily registrations of events and activities is a means to avoid misunderstandings between parents and staff, and to support the aim of participation. Such registrations do not have to require much effort from the staff

Weekly plan							
Time	Monday	Tuesday	Wednesday	Thursday	Friday	Sat.	Sunday
07.00	Wake up, medication						
07.10	Hygiene, dressing						
07.45	Breakfast						
08.15	Dressing for work						
08.30	Transport to work						
14.45	Return from work						
15.00	Dinner						
15.30	Domestic activities (act. bank)						
16.30	Physical activity (act. bank)						
19.30	Supper						
20.00	Leisure time (act. bank)						
20.45	Undress and hygiene						
21.30	Good night						

Figure 23.6 A Week Plan

Registration of Participation in Activities																																	
Name: Erik Jenssen		Month and Year: October 2018																															
No.	Activities	Date:	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	24	25	26	27	28	29	30	31
1	Play Cards	✓							✓	✓	✓						✓	✓										✓	✓	✓	✓	✓	✓
2	Play Board Games				✓				✓	✓	✓			✓	✓	✓						✓	✓	✓	✓			✓	✓	✓	✓	✓	
3	Play Piano																																✓
4	PC games				✓			✓		✓	✓							✓				✓	✓									✓	
5	Listen to Music	✓				✓																			✓							✓	
6	Check/write E-mails								✓	✓	✓							✓													✓	✓	
7	Walking tour									✓	✓														✓							✓	
8	Physiotherapy program											✓				✓					✓		✓		✓	✓						✓	
9	Visiting the gym	✓			✓					✓	✓											✓		✓								✓	
10	Mountaineering wall								✓	✓	✓																				✓	✓	
11	Relaxation exercises			✓		✓												✓														✓	
12	Stretching exercises in bed				✓							✓						✓					✓		✓							✓	
13	Concerts										✓	✓										✓	✓	✓							✓	✓	
14	Café										✓	✓										✓	✓	✓							✓	✓	
15	Visit sister Ida	✓								✓	✓																					✓	✓
16	Visit Parents			✓	✓							✓						✓				✓					✓	✓	✓	✓	✓	✓	✓
17	Winter camps					✓	✓				✓												✓	✓		✓	✓					✓	✓
18	Set the table									✓	✓												✓	✓	✓							✓	✓
19	Cut the vegetables									✓	✓							✓				✓	✓	✓		✓	✓					✓	✓
20	Empty the garbage bin				✓	✓																										✓	✓
21	Go shopping			✓					✓	✓	✓													✓		✓						✓	✓
22	Sort screws and nuts	✓						✓	✓	✓	✓		✓					✓					✓		✓	✓						✓	✓
23	Knitting machine	✓																					✓									✓	✓
24	Empty paper bins					✓					✓											✓	✓	✓								✓	✓
25	Make candlesticks	✓					✓	✓	✓	✓													✓	✓	✓							✓	✓
26	Fill paper - copying machine	✓				✓	✓	✓	✓			✓										✓	✓	✓		✓	✓					✓	✓
27	Initials (maximum 2 letters):	AG	ØE	ØE	ØE	ØE	SR	SR	SR	ØE	SR	ST	ST	ST	SR	AG	ØE	AG	ØE	AG	AG	AG	SR	ØE	ST	ST	ST	AG	AG	AG	SR	SR	AG

Figure 23.7 Form for registration of participation in activities

and should be based on the quality of life issues collected from the *Activity Bank* and the *Annual Wheel*. Electronic registration may make the registration easy to maintain (Figure 23.7).

Figure 23.7 shows a registration form dated October 2018. Similar registration forms can be used for evaluations, for instance to explain why certain activities were not taking place according to plan. It may also be used for communication between parents and staff. Comments in the present study indicate that some staff members favor certain activities, and that the personality or interests of staff members will influence the daily programs of young adults with JNCL. This may not be wrong, but it is important that such patterns are made explicit and discussed. Daily registration of activities and participation may help to disclose such patterns.

Tool 6 – The Log Book

Residential staff, staff at work and parents should be required to post daily entries into a log book. The log book should contain information that is not covered by other forms of registration, for instance information about health, sleeping patterns, or unexpected changes in the daily program. The log book follows the person between residence, work and the family home, and is updated on a daily basis.

The local government may have an intranet for all their service providers and staff, where exceptional situations are noted, and to which parents may have (limited) access. Despite this, parents find that there is still a need for a handwritten log book. It is a personal, less formal and often more useful instrument, according to the parents. The log book can also be used to promote targeted dialogues and communication with the person with JNCL, for instance discussions about the events or happenings that took place last weekend with friends or the family.

Conclusions

It is possible for individuals with JNCL to lead a full life after reaching adulthood. However, given the circumstances, it is necessary to start planning the transition to adult life early, and to involve educational authorities and service providers in this project from the very beginning. Assistance and advice should be solicited from competent sources, which certainly will include parent organizations, local and regional authorities, and national competence centers where these exist. Maintaining and upholding the best possible quality of life for the young adult with JNCL require tight and effective collaboration between family, staff and the person with JNCL. There should be regular meetings between the family and the staff at the work place and in the residence.

The Habilitation Plan should be used for planning and implementing the transition to adult life. It should have a strong focus on participation in activities. It should be supplemented with tools like the Book about (name of the person), the Annual Wheel, the Activity Bank and the Week Plan. Daily registrations of what the young adult with JNCL has participated in may promote a good working culture among staff members and contribute to an optimal quality of life for the young adult with JNCL. In addition, parents find the informal Log Book to be an excellent tool to maintain effective communication between the family, service providers and staff.

References

- Bricout, J. C., & Gray, D. B. (2006). Community receptivity: The ecology of disabled persons' participation in the physical, political and social environments. *Scandinavian Journal of Disability Research*, 8, 1–21.
- Gray, D. B., & Hollingsworth, H. (1999). Development of new measurement tools for studying participation in context. Presented at American Public Health Association Annual Meeting, Chicago, Illinois, November, 1999.

- Krusche, S., Seitz, A., Börstler, J., & Bruegge, B. (2017). Interactive learning: Increasing student participation through shorter exercise cycles. In *Proceedings of the Nineteenth Australasian Computing Education Conference, January 31st to February 3rd 2017* (pp. 17–26). New York: Association for Computing Machinery.
- Magasi, S., Wong, A., Gray, D. B., Hammel, J., Baum, C., Wang, C. C., & Heinemann, A. W. (2015). Theoretical foundations for the measurement of environmental factors and their impact on participation among people with disabilities. *Archives of Physical Medicine and Rehabilitation*, 96, 569–577.
- Oakley, P., & Marsden, D. (1984). *Approaches to participation in rural development*. Geneva, CH: International Labour Office.
- Skye, E. P., Wagenschutz, H., Steiger, J. A., & Kumagai, A. K. (2014). Use of interactive theater and role play to develop medical students' skills in breaking bad news. *Journal of Cancer Education*, 29, 704–708.
- Stanford University School of Medicine (2018). Interactive learning. Downloaded from <http://med.stanford.edu/smili/interactive-learning/>
- Stark, S., Hollingsworth, H. H., Morgan, K. A., & Gray, D. B. (2007). Development of a measure of receptivity of the physical environment. *Disability and Rehabilitation*, 29, 123–137.

24

The Diagnostic Process and Parental Needs and Support

*Svein Rokne, Bengt Elmerskog, Merete Staureby, Susan Fugger,
Anne Hamann and Susanne Joensen*

Diagnosis implies the identification of a pathological condition. For parents it can be a process of shock and trauma when the diagnosis of their child is severe (Holliday, Stanley, Fodstad, & Minshawi, 2016). The family is vulnerable around the time they receive the diagnosis. Most children with a recent diagnosis of JNCL are not aware of their diagnosis, but the responses to questionnaires and interviews in the present project (see Appendix A) indicate that the children with JNCL and their siblings were affected by the new situation, by the changes in behavior and the strange events taking place. It is therefore important that there is support from competent professionals and, when possible, also from parent organizations or individual parents who have been through the same situation. It is vital that the process and the support given are perceived as supportive and useful by the family.

A typical story

The following story by a parent illustrates a diagnostic process that worked quite well.

Our son, who has JNCL, is 29 years old when this is written. We received this diagnosis some twenty years ago and have thus had ample time to think about the way we received these news and what happened during the process of medical investigation leading up to that moment in time.

We discovered quite early that something was wrong with his eyesight, and glasses were prescribed. They did not seem to help much and I remember quite clearly the day on a beach somewhere in Greece when our son, then eight years old, called our names, apparently not being able to locate us five meters away.

On further investigation at the hospital, his eyes were examined and blood samples taken. The blood sample disappeared and he was given the diagnosis of Retinitis Pigmentosa. Apparently this is (or was) often the "first" diagnosis given in these cases. Although this was a shock in itself, we insisted that blood samples be taken again. After several new examinations we were asked to a meeting where we were told the bad news. It must be said that this took place in adequate surroundings and with people (an ophthalmologist and a psychiatrist) who understood their jobs. We were subsequently able to have the diagnosis confirmed by DNA analysis through the good offices of Pirkko Santavuori (1933–2004) in Finland.

Parents are just as different as their children; some become extremely withdrawn and try to come to grips with their new situation on their own, while others seek help. Again fortunately, and unknown to us, there existed an organization which had been entrusted with a national responsibility to support the education of children and young people with this diagnosis and their families. Tambartun Resource Center immediately took charge of a number of practical matters which needed to be dealt with in an orderly fashion and which we, as parents, had no way of taking care of on our own, being at the bottom of the pit, as it were. We did, however, consult the Internet, but this did not give us much comfort under the circumstances.

Tambartun arranged meetings with the local school, they wrote the necessary applications for various technical aids, and we were persuaded that our son now needed to learn braille and that he had certain rights in terms of extra school time to learn this new language. We were also told that there existed a parent organization where we would find other parents in the same situation as ourselves. It must be said that in those days, this institution interpreted their mandate in the broadest possible terms, rendering a number of services not necessarily exclusively within the pedagogical area.

After a couple of years on our own, we finally contacted the parent organization and were able to discuss our situation with peers. This was a very positive experience, and membership in this organization has given us both much in emotional and practical terms over the years.

In retrospect it is clear that we were luckier than many others in our situation. In those days there were no clear routines, and in particular how the diagnosis was conveyed varied considerably. Finally, in 2011, the parent organization in collaboration with resource centers and medical institutions was able to create guidelines and a clear routine to be followed for all new cases. Much had happened in the meantime, of course, including the establishment of a national medical resource center. It has the responsibility

for children and adults with an NCL disease throughout Norway and for coordinating a group of institutions to support all people with NCL and their families. Information to this effect has been distributed to ophthalmologists and others in the medical field throughout Norway.

Requirements for guidelines for the diagnostic process

Several countries have developed guidelines for the diagnostic process, that is, the process from when an NCL disease is suspected until the diagnosis is determined and the family is in a stable situation. Guidelines that follow the requirements for good practice can go a long way towards providing a safe environment for new parents.

Good practice guidelines for the diagnostic process imply that there is a cooperative effort across service providers representing several disciplines, including medicine, education and social work. The guidelines spell out clearly the areas of responsibility and how communication should take place between the various parties.

The guidelines should define four phases: 1) Suspected NCL diagnosis, 2) Determination of the diagnosis, 3) Provision of information about the diagnosis, and 4) Immediate support phase. The guidelines should spell out clearly who is responsible, precisely what needs to be done and how it should be done. The aim of the guidelines is to ensure consistent best practice in relaying the diagnosis to the family and the immediate follow-up for the family. This should include the presence of representatives from the parent organization when possible, who will talk with the parents when and if it is desired by the parents.

The immediate support phase is completed when a functioning municipal support service for the family has been established and a Responsibility Group (see Chapter 11) for establishing and maintaining the individual plan or habilitation plan is in place.

Guidelines following good practice principles have been established (and are being followed) in several countries, including Denmark and Norway. The guidelines may be obtained from the national parent organizations or from the centers that are responsible for providing services to children and adults with NCL, and may serve as a template for providing good practice locally.

The guidelines practiced in Denmark today are presented below. It should be noted, however, that consideration should be given to local conditions when preparing guidelines in other countries.

Danish guidelines when a child is found to have JNCL

In Denmark, the municipalities are responsible for services for people with additional support needs, including persons with disabilities or a rare disease. The social welfare system allows families to choose either to keep their children or young adults with JNCL at home, or in an apartment or sheltered house provided by the municipality. In both cases, the young adults with JNCL will get the help and support needed, and they receive a social pension from the age of 18. At present, most young adults with JNCL in Denmark have their own apartment connected to their parents' home. For support and care, the municipality provides support and care, and teachers and nursing staff come to the home as well as to the school(s) or activity center.

The Danish NCL Team (The Team) is organized and financed by the government, and is part of a national institution for the visually impaired. The support and advice provided by The Team *supplement* the services provided by the municipality.

Guidelines for relaying information about the diagnosis

These guidelines are a highly valued tool for The Team, as they are covering maybe the most important part of The Team's work. The guidelines outline best practice(s) in supporting parents in a situation of severe shock after having received the message that their child has JNCL.

When a child has been diagnosed with JNCL at the Medical Genetics Laboratory of the Danish Kennedy Center or a local hospital, the coordinator or the medical consultant of The Team will be contacted in order to find a time and place for a meeting with the parents. It is very important that The Team is contacted *before* the doctor informs the parents of the diagnosis. The Team will contribute to arranging this meeting in the best possible way. The doctor is advised, in advance of the meeting, to inform the parents that they are called in for a serious conversation and advise them not to bring the child or any siblings.

Two members of The Team – preferably one pedagogical consultant and a social counselor – will be at the hospital on the day the parents receive information about their child's diagnosis, and will join the meeting after the doctor has informed the parents. The Team will have a short preliminary meeting with the doctor, in preparation of the doctor's meeting with the parents. The purpose of The Team's presence is to offer help in a chaotic situation and inform the parents of The Team's existence and that assistance from The Team is available from this moment.

According to the guidelines, the doctor informs the parents of the child's diagnosis, and then the two members of The Team are invited to join the meeting. At the meeting, the two representatives of The Team tell the parents about The Team, that the function of The Team is to support the family in different ways. In addition to a brief presentation of the work of The Team, and equally important, the parents are informed about NCL Danmark (The Danish Parent Association) and that the two parent members of The Team are prepared to meet parents if and when the parents wish to do so.

The Team offers to visit the family as soon as possible, preferably the next day. A declaration of consent form is brought to one of the first meetings with the parents, allowing The Team to inform the family's municipality, school, and other services when needed. The Team offers to drive the parents home in the parents' own car after the diagnosis meeting.

Support from the professionals in The Team

The parents will be shocked after the diagnosis meeting, and it is therefore of importance that they are supported without being overwhelmed. Over time, The Team will talk with the parents about present needs, who they should involve, and future needs. These conversations take place in a way and at a pace that the parents can cope with. The situation of the family has changed dramatically, but the child's life situation at this time is not changed as a result of the diagnosis.

The Team offers to participate in a meeting with relatives and friends of the family to inform them about JNCL and answer the many questions that arise from people close to the family. The parents decide if and when they want to have a meeting, and who they want to include.

The parents often wish to see the medical consultant of The Team, meaning the professor and chief physician at the Center of Rare Diseases, where all the children with JNCL receive continuous follow-up. At this stage, the medical consultant can give insights into the disease, whereas later with the progression of the disease, the medical consultant will be closely involved with the parents in relation to the medical treatment of symptoms and other medical issues.

As soon as the parents are ready, the coordinator and the pedagogical consultant of The Team will contact the local municipality in order to inform the school, kindergarten, etc. of the diagnosis, and of the present and future needs of the child. The Team recommends the creation of a network of professionals of the municipality: social workers, teachers, pedagogues, visual consultants, and so forth. Both The Team and the parents participate in these meetings.

Support from NCL Danmark

With the parents' approval, all the members of The Team are informed that a new child has received a JNCL diagnosis. It varies how early the parents want to meet other parents. However, the message from the parents in The Team is clear: «Do not hesitate to contact us».

The leader of NCL Danmark (Danish NCL Family Association) is also informed that a child has been newly diagnosed with JNCL (see Chapter 26). Also the other members of the Association are informed, possibly without mentioning names, if the parents give their consent. At the diagnosis meeting, the parents are informed about the Association and its activities and services for children and parents. They are also told that one purpose of meeting NCL Danmark is to learn from other parents in same situation, that is, from parental experiences based on «learning by doing». These procedures have generally been highly valued by parents of children with JNCL in Denmark.

How are parents supported?

Parents have considerable needs for support in addition to medical support. The family needs to know how to cope with the needs of their child but also teachers and local authorities need information as soon as possible after the child has been diagnosed. The competence needed is in most countries represented by parent organizations and competence centers. Usually, neither the teachers nor the local government have previous knowledge of JNCL.

The parents in the present project (Appendix A) were asked to consider who gave useful information during the first year after the diagnosis was confirmed. Figure 24.1 shows that parents found that other parents and parent organizations gave most useful information, together with competence centers. In addition, books, articles and the Internet were also considered good sources of information. Parents in other studies have also mentioned the Internet (Cozart et al., 2017). The health sector was expected to be an important source of useful information but the results for the present project may indicate that information from the health sector was not useful in helping parents to meet the challenges of daily life. Medical information about JNCL is important but tends to be more about declines than what can be done to support the child with JNCL in the best possible way. The municipality or other local authorities, such as educational or psychological agencies and schools, were regarded as giving little useful information (Figure 24.1).

Typical comments from parents in the questionnaires or interviews are that local doctors, physiotherapists and the municipality do not have sufficient

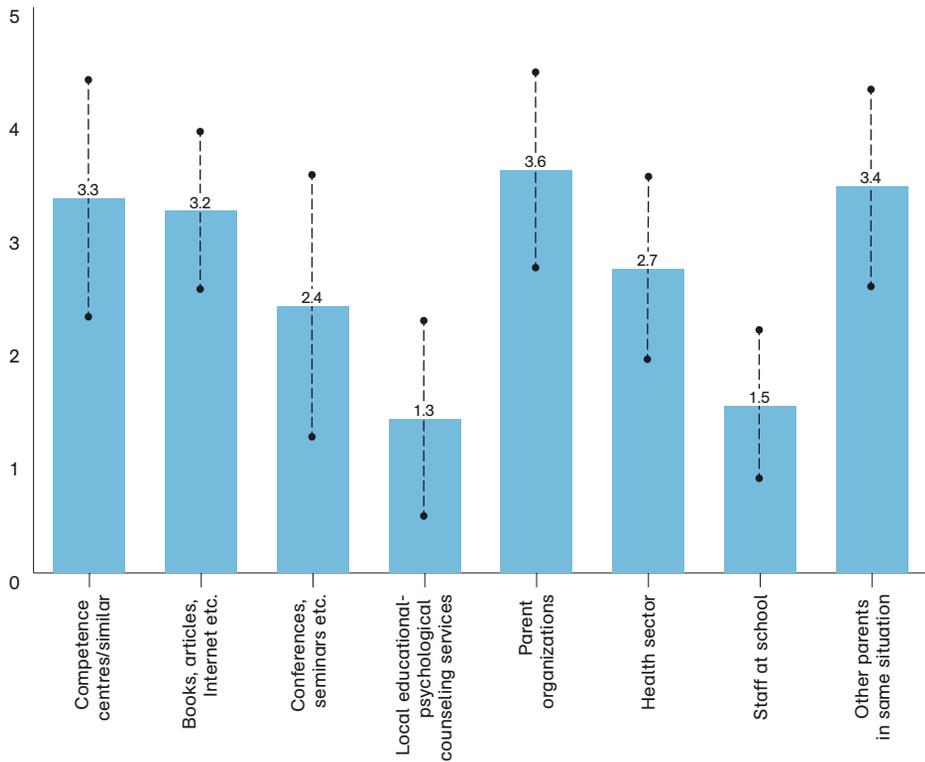


Figure 24.1 Parents' evaluations of information from different sources; average scores on a scale from 1 (no contribution) to 5 (very high contribution), and standard deviations

knowledge about JNCL to provide good services. The contact between the family and the health sector tended to be rather limited in the early phase of the disease. Many parents said that they had to "clear the road themselves," sometimes in collaboration with resource centers and parent organizations. Specialist organizations such as BDSRA in the USA, Onerva in Finland, BDFa in UK, The NCL Team in Denmark, BZBS in Hamburg and Statped in Norway were mentioned as giving useful information. Some parents emphasized that competence and usefulness of information provided were related to individuals in the system rather than the system itself. In fact, some parents considered local authorities more as a burden than as a support.

It is often stated that parents of children who have recently received a severe diagnosis are not receptive to information related to education, development and learning. However, comments in the questionnaires and interviews in the present project emphasized the following urgent need. – «What can I do to help my child in best possible way?» There is always a risk that preconceived assumptions may

preclude proper action by professionals, for example they may give less information because they believe the parents are not ready for it. Schools and other institutions should provide services in accordance with the needs of the individuals. The results from the survey emphasize the importance of implementing tailored initiatives as soon as possible after the diagnosis has been established.

The Danish system illustrates that it is possible to establish an efficient system for managing early interventions and immediate measures. The Team, with different professionals and parent representatives, has been given a strong mandate to contact and follow up families when a child is diagnosed with JNCL and support authorities and institutions in the municipality when needed. The Team is given resources to support all such families in Denmark. The fact that The Team consists of professionals from the health sector as well as other services makes it possible to act fast when needed. Feedback from the Danish families indicates that they are satisfied with the support they receive from The Team.

One important finding in the present project is that specialists within the health services and specialized educational services, as well as parent organizations, are the key sources of useful information for the families and schools. These key representatives can be found in all the countries participating in the project, however, they vary in availability and in how they can coordinate their support. All the Scandinavian countries have a registration system and a service structure which ensure that all children with JNCL and their schools sooner or later will be contacted by the support systems. The systems are different in the USA, UK and Germany. These differences may be related to variation in population and geography but also in national laws and regulations.

Denmark has developed a model for effective accessibility. Also Finland and Norway have established systems where families and schools sooner or later will be offered competent support. However, the coordination or collaboration between the health services, educational services and social services in these countries is not always as efficient as could be desired.

The immediate support phase

Some parents may not be able to act appropriately just after the child has been diagnosed with JNCL. Life may be filled with grief, shock and trauma. This is a phase where parents might need a support system that can act proactively on their behalf. The main issue in the immediate support phase is the child's schooling. A well-functioning school is an effective means of supporting the families.

From the child's perspective, the school situation is not different immediately after the JNCL diagnosis is established, provided the education accommodates the child's daily needs at school. However, there is an immediate need for *building competence* in staff, related to childhood dementia and especially to precautionary learning and hastened learning (see Chapter 12). Parents may not be able to be involved in such interventions at the beginning, but it is necessary that parents are informed about the actions that are taken to promote the best possible development in a difficult situation. At this time, it is also important to establish a close collaboration between the school and institutions that have the required competence about education for children and young people with JNCL. This should be handled as a matter of urgency – there is no time for waiting. The questionnaire comments and interviews of the present project show that many parents highly appreciated the presence of early proactive actions taken on the child's and the family's behalf until the family situation had become more stable. However, they also show that many parents felt very lonely in the beginning and lacked the immediate support from the community and the education system. In the words of one parent:

There were a lot of people involved in the beginning which was very overwhelming. I don't really know how school was supported. I think the school tried to find information. I did very often mention the BDEA but that was never taken up. Maybe the RNIB was contacted.

This is no criticism really of the school. They couldn't – they didn't really allow my child to go out into the playground and play with the other children. Because focus, obviously health and safety and – and I understand it. They – they wanted to make sure she was safe. So that became hard and difficult for them – and us.

I felt I couldn't trust the teachers and their competence. In my opinion, the teachers were not able to deal with the illness and did not have sufficient expertise. They covered a basic program, but it was me who supplied my daughter with additional materials. For instance, I prepared workbooks or exercises from the early beginning.

This short comment emphasizes the need for both information and action, for parent support as well as dialogue and participation. The function of the guidelines is to provide a foundation for this.

References

- Cozart, E. J., Augustine, E. F., Mink, J. W., Thatcher, A. R., & Adams, H. R. (2017). Batten disease and parents: Marital quality, support, and communication. *Journal of the International Child Neurology Association, 17*, 114.
- Helland, I. B., Barlinn, J., Åbyholm, G., Paus, T., & Rokne, S. (2016). *Guide to symptomatic treatment of neuronal ceroid lipofuscinosis*. Oslo: Norsk Spielmeier-Vogt Forening and Oslo University Hospital.
- Holliday, E. L., Stanley, H. C., Fodstad, J. C., & Minshawi, N. F. (2016). Stress and satisfaction in the diagnostic process. In J. L. Matson (Ed.), *Handbook of assessment and diagnosis of autism spectrum disorder* (pp. 137–155). Cham, Switzerland: Springer.
- Labbé, E. E., Lopez, I., Murphy, L., & O'Brien, C. (2002). Optimism and psychosocial functioning in caring for children with Battens and other neurological diseases. *Psychological Reports, 90*, 1129–1135.
- NCL Danmark (The Danish NCL Family Association). <http://dsvf.dk>

Being the Brother or Sister of a Child with Juvenile Neuronal Ceroid Lipofuscinosis

*Minna Sarola, Egil Rian, Linn Sophie Rian,
Charlotte Holmen and Ida Holmen*

The family is a social system, where the members influence one another. Children spend much time with their siblings, often more than with their parents (Howe & Recchia, 2014). Sibling relationships represent both social resources and challenges, and the family's emotional climate affects the relationship between siblings (McHale et al., 2012). Sibling relationships are usually based on some sense of being equals, even though age differences can lead to a number of differences in status and physical strength. When one sibling has a disability, this may change the relationship in some ways (Meltzer & Kramer, 2016). Children with severe disabilities often become "younger siblings", regardless of their actual age ranking in the family. In many areas, the relationship is the same as in siblings without disabilities but characterized by more warmth and care than other sibling relationships (von Tetzchner, 2019). For severely disabled children, who may have difficulties engaging in ordinary peer relations, siblings typically have a very special role (Petalas et al., 2015).

This chapter presents experiences of being a sibling to a child or young person with juvenile neuronal ceroid lipofuscinosis (JNCL). It includes results from a study where siblings of six individuals with JNCL were interviewed (Sarola, 2016), a sister's and a mother's account of the sibling relationship, and a father's interview with his daughter whose brother had JNCL.

Six families' views on sibling relations

A child's illness or disability is always hard for parents and when a family member falls ill, it affects the whole family (Mäenpää, 2014; Seligman & Darling, 2007). It may absorb many of the parents' resources and influence the life of the entire family.

When he was at home there were a lot of things to take care of, like feeding him and helping him to go to the toilet. It made everyday life much harder but we survived pretty well.

We had ramps installed in our house and everything you need when you use a wheelchair.

When a child in the family is diagnosed with JNCL, it therefore affects the life of the siblings. Anxiety, fear, uncertainty and mourning of functional loss become a part of everyday life (Kaulio & Svennevig, 2008). Much of the time and energy of the parents may be focused on the child who is ill, and the siblings may get less attention. In particular when the child with JNCL experiences health problems or needs hospitalization, the parents' thoughts may be pre-occupied with the treatment of the child. This inevitably affects the life of the siblings (Kulomäki, 1984). Parents and professionals therefore need to be mindful of the siblings and their need for support (Sarola, 2016).

Siblings may also feel that after their brother or sister has been diagnosed with JNCL, there are greater expectations on their own shoulders. This can cause considerable pressure, both at school and at home (Kulomäki, 1984). Sometimes siblings of children who are ill feel the need to protect their parents from any extra stress, and therefore hide their grief (Kaulio & Svennevig, 2008). Older siblings may take on a caregiver role and take great responsibility for the sibling who is ill, as well as for everyday household tasks. Moreover, siblings may feel that this is the only way to get their parents' attention and acceptance (Kulomäki, 1984). However, under a surface of strength and kindness there may be hidden feelings of anxiety or depression (Kaulio & Svennevig, 2008). One of the siblings in the interview study described the situation:

One year after the diagnosis, it started to show in me. I was depressed and I suffered from an eating disorder. It happened because I hid my own problems and tried to be as good as I could be to make my parents' life a bit easier. At school I only got top marks.

The interviews confirmed that the illness of the child was a permanent part of the everyday life of the siblings. In fact, the life of the family was to a large extent determined by the nature of the illness of the child. When an illness is progressive like JNCL, the family has to adapt to new situations continuously. All the siblings mentioned that the parents gave more time to the child with JNCL, and that they had to some extent concealed their own problems and worries to make their parents' life a bit easier. The siblings also found it necessary to take

more responsibility for themselves than a child of their age would normally do. Adults should always remember that there may be more going on than the sibling might show.

Disease evokes feelings

The attention given to a child who is ill may cause envy in siblings (Kulomäki, 1984). A child's disability can raise a variety of emotions in siblings, such as anger, joy, confusion, fear, love and pride (Finnish Association for Parents of Children with Visual Impairments, 2015). Siblings may also feel guilty because they are healthy while their sibling is ill (Batten Disease Family Association, 2015), as this sibling explains:

Sure I thought about why I get to be healthy while my sister is ill. When you see your sister suffering, it makes you wonder what right you have to be healthy.

The way parents respond to the disease will also influence the reactions of siblings (Kulomäki, 1984; Trachtenberg & Batshaw, 1997). The child who is ill is a large part of a sibling's life and the situation may make them more open minded and also affect their identity formation. Siblings may wonder what their life would be like if the child did not have a disability and how different it would be if the child was not ill (Kulomäki, 1984). Allowing all kinds of feelings and talking openly within the family may help siblings adapt to an extraordinarily difficult situation (Kulomäki, 1984).

I felt my parents' sadness even though they tried to cover it up.

At times I was ashamed of my brother even though I thought it was wrong to feel that way. But as a child, illness sparked all kinds of emotions.

I'm not bitter towards my parents even though most of their energies went on my sisters, but still I feel that I was little bit of an outsider in the family.

All the interviewees felt that it was natural for a sibling to experience a wide variety of feelings. They also said that it was difficult to witness their parents' grief. The interviewees felt that it was important for a sibling to feel that all possible emotions are natural and permitted. They also considered it very important to have opportunities to discuss the emotions that the disease had evoked in them, either with their parents or with another adult.

The need for information

All the interviewees expressed the view that siblings have a right to know about the diagnosis. They all felt strongly that factual and age-appropriate information had helped them understand the disease and its symptoms. The siblings interviewed also pointed out that children are very sensitive and notice if their parents are not coping. There is an abundance of inaccurate information easily accessible on the Internet. All the interviewees said that a lack of knowledge or incorrect knowledge is much more harmful than factual and reliable information about the diagnosis.

When you have enough information, you are able to understand that the symptoms are just part of the disease and that makes it easier to handle.

I was told about the disease little by little. I was around ten years old when I heard where it would lead in the end.

The fact that JNCL is a hereditary condition also raised some important questions and the siblings had just as many thoughts and questions as the other members of the family (Norio Centre, 2015). Factual and age-appropriate information is important when the illness of a child raises questions. Siblings need to know why the child who is ill needs so much attention and care (Kulomäki, 1984). A rule of thumb is to answer any questions that the sibling is asking about the child's illness honestly (Vehmanen & Vesa, 2012). However, it should be borne in mind that a sibling may not know how to ask all the relevant questions. For example, if the parents do not talk about JNCL but only about visual impairment, a sibling may not be able to ask questions about JNCL, and may therefore depend on other accessible but misleading information (Parisaari, 2015). One of the siblings in the study expressed this view.

If you sort of know in advance that the ability to speak and move are going to get worse you are able to better appreciate the times when those skills are still there, and you don't feel like you wish you had known it was going to get this bad so you could have enjoyed the moments when she was less affected.

Children under school age are often open-minded towards diversity, and at this age a sibling's disability does not necessarily raise any particular thoughts. In general, school-age children begin to reflect on these matters: Why is my sibling different? Am I different? Why do some people have disabilities? What causes disabilities? At this point, when the questions are topical, it is essential that the

relevant information is to hand. Incorrect perceptions can cause anxiety and fear. For example, a sibling may believe that he has caused the child's disability if he does not have correct information about it (Vehmanen, 2012). Moreover, it is important for siblings' self-esteem and development that they know how to tell their friends about the disability of their brother or sister (Vehmanen, 2012).

Peer support

All interviewees said that support from peers was vital. Some of them had experienced peer support while others wished that they had received more. They all said that it was a relief and empowering to meet others in the same situation. It was comforting for them to realize that there were other families in the world experiencing a similar situation.

A peer group can share common traits such as age, gender, culture, religion or ethnicity, shared experience, sexual identity, health or education (Rubin et al., 2015), and many support programs focus on peer relations (Western Australian Centre for Health Promotion Research, 2015). The idea of peer support is to give and receive help by sharing experiences (Inclusion Finland KVTL, 2015; Repper, 2013). Peer support and professional help are two different things. Sometimes a peer can offer the best help to another person (Kinanen, 2009; Repper, 2013).

I would have liked to have met other families in the same situation. Sometimes it felt like we were the one and only family like this and nobody else could understand what it was like and how we felt.

The interviewees said that meeting other families was also important because they saw other children who were at different stages of JNCL, even if it was hard to see children where the disease had progressed further than in the child in their own family. This was important because seeing how the disease would progress helped them prepare and adapt to the idea of their brother or sister getting worse. The interviewees said that it made them appreciate the time when the child's general condition and mobility were still good. The siblings said that seeing other families with a child at a different stage of the disease progression evoked feelings and questions which had been important to talk about with an adult. Seeing how other families had managed their situation gave them comfort and hope for the future.

I remember that it was frightening at first to see how the disease had progressed in children of other families. On the other hand, it was very important because in the future your own sibling's disease would progress too, and seeing others was a way to prepare yourself for it.

In conclusion, the results of the interview study indicate that adequate information and peer support have significant value to the sibling's ability to live happily as part of a family where a child has JNCL. Kinnunen (2006) found that peer support has a positive effect on parents of children with special needs. Peer support is seen as hugely beneficial when, for example, dealing with the feelings that a child's special needs evoke in parents (Kinnunen, 2006). Kolehmainen and Pulkkinen (2009) found that sisters of those with an intellectual disability felt that the peer support that they had received was empowering and useful, and those who had not had peer support felt the lack of it. Their comments support the findings of this study.

Reflections of a sister and a mother

This subchapter consists of two independent narratives about a young man, Nils aged 25 years, with JNCL. The first is written by the young man's sister, Ida, when she was 23 years old. The second is written by the young man's mother.

Being the Sister of my Brother

It is not easy growing up as the sibling of a child with special needs, but with great challenges come great rewards. I could not have asked for a better family and I am forever grateful for the joy my brother gives me and for the love my parents radiate towards me. But it was a bit harder for the blond, shy, little girl to understand the great strain that had been put on her parents when she was just a child. I was three years old and my brother Nils was five when he was diagnosed with JNCL. At that moment, the dynamics of our whole family changed.

The different adaptations and coping mechanisms I developed during those early years have become a large part of who I am today. My parents time and energy were, by necessity, divided unequally between me and my brother. I adapted to the fact that my brother always took the bigger piece of the cake, so I convinced myself that the smaller piece was good enough for me. I felt my parents' grief, despair, and stress, so I tried as hard as I could to make them happy, and to unload their stress. I understood that my parents had enough to worry about, so I stayed out of trouble and became a perfectionist and peacemaker. In all of this "rightfulness", I lost myself. I did not realize this until I moved away from home, and found myself at the age of nineteen with no clue about my own needs and wants, not even what I preferred to eat for dinner. I had a hard time making decisions, even with no-one else to consider in the equation. I only had to focus on my own desires and this paralyzed me. I was so used to choosing whatever was best in a situation by



Ida and Nils 20 years ago.

calculating all the different variables that I forgot to pay attention to what I wanted. I guess knowing what you want is also a part of growing up, so it is not only my role in the family that got me to that point. I still have some problems separating what I want and what I should want, but as I get older and more confident, I am also learning more about my own needs and desires.

It is important that the sibling of a person with special needs feels seen and heard. It is easy for an adult to forget that the three-year-old sitting on their lap and smiling also is feeling what is happening to the family. Even though it is totally understandable that the person who is ill gets more attention, it is important to be aware of the impact of this on the sibling. I have always felt the enormous love my parents have for me, and I know that they have done their best for both of us, and that is all they can do. I am lucky and truly thankful for the childhood I have had. Lucky in the sense that my family is resourceful and we have been able to create a good life from what has been given to us. Luck should not be a variable in the debate of outcomes. All families should be given the possibilities we have, and everyone should know their rights.

I have now lived in the United States for a couple of years, and the hardest part is being away from my family. We are very close and a great team together, so it is difficult for me to feel whole without them. I feel both beautiful joy and dark pain every time I come home after several months away. The beautiful love



Ida and Nils today.

was eighteen. It would have been hard to understand the fact that my brother, most likely, will not always be by my side. It is still hard to understand. I chose not to do any research about the disease the first years after I heard its name. I was, and still am, confident that my brother has a new form of JNCL, so I figured I would not bury myself in the dark truth if there is a chance that it does not apply to him. Since we have been advised to protect my brother from knowing the outcome, we have chosen to keep the severity of the disease from our community. Growing up with a big family secret also shapes you. None of my friends really knew what I was going through and how serious my situation actually was.

When I look back, it was hard growing up faster than most of my peers, but I think it has only made me stronger. I am good at keeping secrets and have

we have for one other flourishes, but the severity of the disease hits me when I can see more clearly that my brother is slowly getting worse each time I am away.

The hardest thing for me to face is that in the future my eccentric food-loving brother will not be able to eat normally, my curious question-machine will not be able to talk, my active nature-loving adventurer will not be able to walk, and I will not grow old with my hero by my side.

At the same time, the ability to eat, talk, walk or live can be taken away from any person at any time. The difference is that my family has already been given a kind of guarantee that it will happen to one of us. I do not want to say that we are lucky that this has happened to us. Absolutely not. But I want to say that we are lucky in the great love our situation has brought us. Love that touches more people around us than we know.

I am glad that I did not hear the whole truth about JNCL until I

become a trustworthy person. I also feel that I have developed a stronger sense of self and what really matters in life. I still remember scenarios from high school where some of the girls were devastated because they got the wrong color phone for Christmas. A feeling of annoyance surged inside me when they could not see the insignificance of such a problem.

Today, the three-year-old me has become twenty-three years old, and I am extremely proud of each member of my family. I am convinced that we would not be filled with so much love and strength without the challenges we have been through. We will experience great grief and sorrow in our lives but we will have each other, and that is all that matters. To be the sister of my brother is the best thing in the world, and I cannot wait to move back home.

Siblings – A mother's reflections

It has not always been easy to be the little sister of Nils. He always demanded a lot of attention from me and his father, and he needed a lot of help. Our daughter Ida retreated and withdrew in most situations, patiently waiting for her turn. Nils dominated my life and his father's life; he was loud and Ida was silent, and he would shout if not receiving enough attention. Ida has never complained, but we observed that she often felt sad. It is difficult to put things into words when so young.

Nils is very proud of his little sister Ida. He has always considered her clever and beautiful. He boasts and brags about her whenever he is given the chance. His loud bragging and boasting about her were at times a bit embarrassing for Ida, but she has never been uncomfortable about being his sister. She was always proud of him and included him in social gatherings, when having friends home or having a party. That he may have looked at bit different, or said weird things, never embarrassed her. She always defended and supported him, irrespective of his appearance and behavior.

Her experience as Nils' sister has affected Ida and her personality. She always defends those with special needs or those who are different in any way. Everyone has the right to be met with dignity. Ida gets angry and despairs when less fortunate people are treated badly, unfairly or without respect. She never judges other people. Her relationship with Nils has made her an open, understanding and helpful person.

Ida was three years old and Nils five when we received the diagnosis at the hospital. Ida was staying with her grandmother and I have always wondered what she thought, felt or understood when we returned home with the diagnosis. She certainly understood that something was wrong. We were sad but we tried our best to keep our spirits up in front of her.

Our lives changed totally after we received Nils' diagnosis. Work and other things in life that had been important before suddenly became insignificant. Other questions took over: Is there really no treatment? Do we just accept the situation? Cancer is treated with chemotherapy and radiation. Can this treatment be used for Nils? Shall we sell everything we own and move abroad to find someone who can help us? Where can we find help? We spent many hours on the Internet looking for answers but there were none to find. What we found and read online was terrible and depressing and it just pushed us further down.

At first we felt panic. We must allow Nils to experience as much as possible before he becomes blind, dependent on a wheelchair and loses his speech. We must travel and allow Nils to see and experience as much as possible before he becomes blind. We must allow him to learn as much as possible before it is too late. But eventually we calmed down. We realized the most important thing for Nils is to feel comfortable with friends and family in his home environment. He needs predictability, security, love and the same challenges as other children his age. He must learn to cope with daily challenges and he must continue to learn new things. Our primary goal was to enable Nils to have the best possible quality of life.

But what about Ida in this chaos? From the very beginning she was a shy and quiet child who did not demand anything. Nils took up much of our daily life. Ida had a more hidden role at home. She quickly became Nils' helper, showing great care and sympathy. Being caring and sympathetic is good in itself, but it worried me that she took too much responsibility. I was afraid that her adaptive role in the family and her selflessness went far beyond her own needs.

I have felt bad because I spent so much time on Nils and less time on Ida. There was always a need for complex planning and organization with Nils. This is a dilemma that I am still living with 20 years later.

Another dilemma I have had through the years concerns Nils "getting everything he is pointing at". Ida has hardly asked for anything and I sometimes feel she has been "forgotten". I remember thinking «it is now we have the opportunity to do things together. In a few years Nils may become so disabled that it is too late. This is our time of opportunity». At the same time I was thinking «we have more time with Ida, it is no hurry». But I soon realized it is wrong to think that way. We live here and now, regardless of any disease, and we have to make the best use of time for us all. Wasted time will not be given back and quality time with Ida is important now and in the future. We are always keen to give a lot of praise and positive feedback to Nils and Ida. I think this is very important, regardless of the situation and despite their abilities.

Early on we focused on establishing an "open house" for Ida and Nils, and their friends were always welcome. There have been lots of indoor and outdoor activities taking place. At times it has been tiring, but we are grateful that we have

been able to give priority to this. I am sure the open home has benefitted both Nils and Ida. They are both very sociable with wide social networks. Nils is now living in his own flat with staff funded by the community. Many of the staff were childhood friends of Ida and Nils, and we can see that those early relationships have a positive effect on their work with Nils today. Their efforts are based more on friendship than paid employment.

The fact that Nils is blind and has epilepsy is of course a difficult issue. However, these challenges are easier to deal with than the ongoing cognitive decline. The prospect of our son and Ida's brother having dementia at a young age and eventually losing his speech is tough for us. Nils does not know about his disease. He is aware of his blindness, epilepsy and that he sometimes has problems finding the right words.

Ida was eighteen years old when she was told the full truth about the disease and what is going to happen to Nils in the future. We took the decision to wait until she was eighteen mainly because we wanted her to have a childhood as normal as possible, without too much worrying and sorrow too early in her life.

What could I have done differently as a mother? I have certainly made mistakes, but at the same time it is important to remember that I have also done a lot of good things. Either way, myself and my husband are only human. I think we have done what was possible in our situation.

Ida has learned a lot from having a brother with JNCL, a learning experience few other siblings will experience. Today, she is strong and wise with a great perspective on human life and she has great empathy and respect for people with special needs. These are good qualities for the rest of her life.

Perspectives on siblings

This account is written by a father and his daughter about their son and brother with JNCL. The young man, Henning, passed away some years ago. It is based on an interview, where the father asks his daughter about her experiences as a sister to her brother with JNCL.

Introduction

A story must start somewhere. This family story started in the winter of 1983 and 1984. At the time we thought we were a completely ordinary family of four: mother Vigdis, father Egil, six-year-old big brother Henning, and little sister Linn Sophie who was barely two years old. An ordinary family with good prospects. Most things happened as we expected in life. We – the adults – had both found

a partner we were happy with, we lived in a nice house, and we had good jobs and nice friends. In addition, we had two beautiful and healthy children. Our lives progressed as we had wished; things looked really promising, with no major problems. Of course.

But our family had reached a turning point. During the winter of 1983–1984, it was discovered that Henning had some vision problems. We thought these problems would be corrected with a pair of glasses or something similar. Henning's vision was assessed by an ophthalmologist. When we returned to the doctor, nothing turned out as we had hoped or expected. The doctor was very worried and immediately referred Henning to hospital for further assessment. We were told that something could be seriously wrong with Henning. In April the same year, Henning was given a diagnosis that hugely affected the life of our entire family. Of course, we had never heard of Spielmeier-Vogt disease or JNCL before. The information the doctor gave us about Henning's future shocked us deeply. Nevertheless, life had to go on. We have tried to cope with the situation the best we can, despite all the challenges we have experienced over 30 years. It has been a very long journey for us all.

Henning lived a long life, considering the typical outcome of the disease: he was 38 years and three days old when he passed away. We do not know anyone else with JNCL who has reached this age. When we, the three remaining members of the family, are sitting together reflecting on our history and where life has led us, we have many memories. Many good and many difficult memories. The sadness of losing Henning still marks our lives, but we are also very grateful for everything we received through Henning and the resulting close family bond. Many have commented that we became a very close and intimate family. A good friend referred to us as the "Rian Club" and we identify closely with this name.

We – the remaining family members – have our mutual experiences and history, but we also each have our own, individual story. What we remember and what we have forgotten are individual matters. The same story can be told in different ways, based on how we process memories. The same story can also be shaped differently, based on individual differences in interests and emotions, and what is of importance to the individual. This is the story of little sister Linn Sophie.

The story of little sister Linn Sophie

FATHER: Linn Sophie, you were not even two years old when Henning got his diagnosis. This means that we – your parents – had to reflect on your behalf on the first challenging years of your life. Even if you did not understand what was happening to Henning and our lives, we could see that you clearly noticed certain things and family challenges. You noticed the change



The Rian Club.

in the spirit and mood of the people surrounding you, and you saw that something was seriously wrong. The way we looked at you, the way we tried to care for you. The situation naturally challenged your need for security and predictability. Our observations did not come from great knowledge of developmental psychology or deep insight into attachment theories; parents can observe how circumstances and conditions are affecting their child. Our family situation had a significant impact on your early childhood.

DAUGHTER: Yes, one might say that. As far as I can remember, I have always been sensitive and aware of the atmosphere and feelings around me. I have been afraid of being alone, afraid of being abandoned. Of course, I remember few specific or concrete events from this time, but my body recalls them somehow. It is reasonable to assume that as parents you were not able to be 100 percent there for me at all times psychologically or physically. You were in shock. In shock from knowing that Henning would undergo a change from being a normal and healthy boy to having a long and sometimes very stressful illness. That it would last for 32 years was absolutely unthinkable at that time. In addition, you did not know initially whether I was healthy or if I would face the same destiny as Henning. For over two years you lived with this uncertainty, before you tested me and found out that I was not in danger. Although I did not know this, I obviously felt something in the atmosphere around me.

I have a daughter now who is three years old. People say she is exactly like me, both in how she looks and the way she behaves. She is very attentive to my emotions, if I am sad or not feeling well. She has given me new insights and understanding about my early life, when you (the parents) underwent the most comprehensive and profound crisis of your life.

FATHER: As time went by, your ability to adapt to ongoing changes in life was always a challenge. You did not have anything to compare to, your situation was so different from others, but you coped in a natural and very impressive way. You were in a situation where you had to develop skills and ways to care and support Henning. You undertook responsibilities unknown to most children of your age. In addition, you had to cope with our worries and strains. We were not able to hide the truth from you, or what would eventually happen to Henning, and what the future would bring.

DAUGHTER: *As I remember it, you started to give me little hints at an early age about what was going to happen to him. Later I understood that I had been fortunate. Fortunate because you included me in your lives and did not put me on the sideline. I feel I have always been an important member of the Rian club. There have always been enough hugs and love in our family. The openness, trust and faith you showed me have strengthened my relationship with you. I feel we are so close, we can talk about anything today. Almost! When I was ten years old, you and Mom told me the truth about Henning's diagnosis. I was aware that he was blind, had epileptic seizures and eventually would be dependent on a wheelchair. One day when Henning was away, we rented a movie called "Lorenzo's Oil". It was about a boy who had a progressive disease. Like most American movies, it had a happy ending. They found a cure for his terminal diagnosis. After we had returned the movie to the video shop, we sat in the car and chatted. I said I was happy that Henning will only be confined to a wheelchair and not die because of his disease. You told me: «In reality the boy in the film did not survive the disease. He eventually died.» The whole truth came out. I remember we were sitting in the car – it was Autumn and it was raining. You told me that the way Henning spoke would eventually become more difficult to understand, and he would probably not live much longer than 20 years of age. What I was told about Henning's time of death was fortunately wrong, but many pieces of the puzzle fell into place, things I had been wondering about.*

The time that followed was tough. I tried to hold my tears back when I was at home, but I cried every day at school. Fortunately, I had long hair so nobody could see me crying. For a long time you tried to establish contact between me and the health system. You wanted me to talk to a professional about things that

were difficult to talk about at home. You did not manage this before my teacher noticed the problem and contacted the local psychological-educational agency. Sadly, I did not get any meaningful support from them. The first psychologist suddenly retired from his job, and I was not able to establish a close relationship with the next one as the chemistry between us just was not there. Neither did you have any close friends or family that could act as support to me. I could not tell anyone what I knew about Henning. Finally I shared the secret with my best friend. She could bear it for a while, but I suppose I became too intense, too cautious, too precious, too close, and too sad. I felt I was the only one in the world with this grief. That nobody understood me. At home, I was no longer the little sister of Henning. I was Henning's big sister. Intellectually I had overtaken him a long time ago.

FATHER: As you became older, Linn Sophie, how did you feel about your life with us, life in a different family? What advantages or limitations were there? We had contact with others in the same situation, but how did you find it?

DAUGHTER: Meeting others in the same situation was very important. My best friend had a big brother who was the same age and had the same diagnosis as Henning. I became very fond of him. Unfortunately, they lived a long way from us, so we could not meet often. We did not talk about sickness and sadness much when we met, but we had a very special relationship because we were both in the same situation. It was good to know that I was not the only one in the world experiencing and facing this.

The job of raising me was certainly not easy for you and I do not feel that you asked that much of me as a sister or daughter. I suppose you felt you could not ask me to do things Henning was unable to do. I finally realized that this was wrong; there had to be more expectation on me than on Henning. Today, as an adult, I think you should have made more demands on me than you did, because I am still a bungler and muddler today!

There were some limitations having a brother like Henning. It was not possible for you to let me participate in all the usual sports and activities. You had more than enough on your plate. It was just not possible for you to do additional things as other parents did, like selling cakes, or tickets for the local handball club or brass band. However, it was possible for me to participate in ballet dance. I loved it, but at the same time I missed being a part of the community that other girls in my class were part of. On the other hand, I experienced many positive things that other children my age never had the opportunity to be a part of. For instance, we went on tours with the family association you were members of,

and on many exciting tours to Mediterranean countries. Resource centers for JNCL, like Frambu (Competence Centre for Rare Diagnoses) and Tambartun (Special Education Centre) also organized meetings. These gatherings gave me great experiences my friends never had the opportunity to have.

FATHER: The fact that Henning lived at home for almost twenty years certainly influenced your formative years. We tried to have an open house, and many children came and went on a daily basis. How did you find this?

DAUGHTER: *When my best friends came to visit me, Henning usually joined in, but sometimes we wanted to be alone without him. Henning could be demanding at times. When he did not feel well, for example, he would sit in his room shouting: «I'm a lonely little blind boy.» I was just not able to compensate for his need to have friends. I would meet up with one of his classmates and ask him if he could visit Henning. Sometimes he said yes. However, the reality was that most of Henning's many so-called good friends were just friends in his imagination.*

Being with Henning was often tiring. His incessant talking was exhausting. He talked about dinosaurs, Indians, Elvis and all the plastic figures he had in his collection. He had hundreds of smurfs, he was particularly fond of the inhabitants of Duckburg (Walt Disney). He talked without drawing breath. At times it was impossible to have a conversation with him; it was always on his terms. You – my parents – could not hide being tired by Henning's behavior. When I saw it getting too much for you, I would take Henning out of the living room, and start reading a book to him. I am told that I did this before I had even learnt to read. We cannot deny that the pressure on us at times was overwhelming. I remember I once drew a painting showing you, Daddy, standing and pointing at the door. Henning was on his way out through the doorway while Mom was crying. When you were very tired, you could be tougher and stricter. This made me afraid that you were ready to have Henning leave our home.

FATHER: As you became a teenager, Henning became more and more affected by the disease and we all had to deal with new challenges. You continued to be a great carer, but you also needed to break away from us, like other young people of your age. Did this raise any issues for you?

DAUGHTER: *I remember I wished that the mysteries behind Spielmeier-Vogt disease (JNCL) would be understood and the problems solved. Maybe there could be a cure for Henning's disease? Maybe in the future I could do medical research and find a treatment or cure? As the years went by, I realized that this was just a pipe dream.*



Sister Linn Sophie and brother Henning.

When I became a teenager, I was still very loyal to you. But I also had a strong need to rebel like other young people. Our closeness meant that you discovered most of the crazy things I did. Well – almost everything! Henning had become psychotic by this time. In his reality, I was no longer his little sister. He was dead, we were all dead, he felt he was losing all his body parts. We had to help him pick up his fingers and put them back in place. There was no smile. No pleasure. Henning was given strong medication and he was "gone" for a long period. After almost two years, the medication was phased out and a wonderful thing happened. Somehow Henning woke up to life again. The psychosis was gone and he became the good, kind version of Henning we had missed so much for so long. But with one main difference – he used far fewer words now. It was sad to see him gradually lose abilities. I do not remember much from this time and I think I was probably more concerned about myself at this age.

I see myself as a positive person, but in a melancholy emotional state. Eventually, I realized that I could not put my life on hold. Nobody knew how long Henning was going to live. When I was 19 years old, I went to Bali to study for three months. I was very worried that something would happen to Henning

while I was away. You, Mom and I had the same feelings and concerns, which caused a bad conscience because I had left him. I tried to compensate for this by sending many sound clips and postcards to you all.

After returning to Norway, I continued to be 400 kilometers from home as I started my nursing training. During my three years of study, I travelled home every other weekend to see Henning. It was important for me to see him because I really wanted to, not because I felt I had to. Then I moved even further away – 1200 kilometres – to Tromsø. The worry continued when I later moved from Tromsø to Oslo, 550 kilometers from home, for further training as a children's nurse. I had particularly difficult times when I thought that Henning must die soon so we would be able to "breathe" again. I loved Henning so much, but to continually worry about the future, without knowing when and how it might end, was just terrible.

Henning and I had a very close relationship. Mom and Henning used to phone me when I was away. Although Henning had lost his speech, he was more talkative than anyone with similar communication issues I have ever met. He spoke to me in his own language, and it was easy for me to translate this into «I love you and I miss you, sister.» These phone calls made my life much easier. But the very best times were when I was at home and could lie in the crook of Henning's arm, and he would be smiling and laughing at having his little sister in his arms.

FATHER: We have often wondered how it was for you growing up with a brother like Henning. What do you think the consequences have been, and how does it affect your life today?

DAUGHTER: Our life with Henning has made me different to other people. I have been aware of this for some time. My opening line when meeting new people was often: «Hello, my name is Linn Sophie and I have a brother who has a rare disease called Spielmeyer-Vogt.» The responses and reactions varied a lot. Some people were curious and asked more. Others just fled. Why was it so important for me to tell them about Henning? It must be allowed to share that he took a lot of space in my daily life. The feeling of being different to others gave me an enormous need to explain my life to them. It is clear that this has scared away some people over the years. I understand this now, but it hurt me every time I felt abandoned by others. I lost many friends because of this. It took time for me to create a distance from these feelings, and to find my own clear identity as Linn Sophie. I now find it easier to share less, but it can still be difficult. Even after his death. I am still so proud of my wonderful brother! Maybe I have been influenced by debates in society about attitudes to people with special needs,

which make me feel I have to explain to everyone how wonderful Henning was. To hear that people like Henning are seen as burdens on society, vegetables, or a waste of money makes me sad and angry. Who would I have been without Henning? Henning shaped me. He made me what I am today. I am very cautious. I am sensitive and vulnerable. But I am also a caring, kind and cheerful person.

Henning's final phase of life was difficult. In retrospect, I am very grateful you involved me in all the care and nursing decisions. At times I had strong opinions about life-extending measures and resuscitation. You allowed me to express my thoughts. Addressing such ethical dilemmas was difficult for you, me and other caregivers who helped him at the end. It was important for me not to be Henning's nurse during this difficult time even though I had professional competence in the field. I just wanted to be his sister.

Henning has taught me to appreciate the small things in life and to realize what is important. Henning is not here anymore but he will never be forgotten. His 38 years long life had a decisive and definitive effect on me and on many others. He really lived his life; he rejoiced in his way of living and lived in a way that many could learn from. His headstone bears the following inscription: «Thank you for everything you taught us about life and love.»

References

- Batten Disease Family Association (2015). Sibling support. Downloaded October 30th 2015, from <http://www.bdfa-uk.org.uk/sibling-support/>
- Blacher, J., Feinfeld, K. A., & Kraemer, B. R. (2007). Supporting families who have children with disabilities. In A. Carr, G. O'Reilly, P. N. Walsh, & J. McEvoy (Eds.), *The handbook of intellectual disability and clinical psychology practice* (pp. 303–335). London, UK: Routledge.
- Finnish Association for Parents of Children with Visual Impairments (2015). Vanhemmat ja perhe ovat sisaruksen tärkein tuki (Parents and family are the most important support for the sibling). Downloaded October 17th 2015, from <http://www.silmatera.fi/nakkarila/elaman-alussa/sisaruus>
- Howe, N., & Recchia, H. (2014). Sibling relations and their impact on children's development. In M. Boivin (Ed.), *Encyclopedia of early childhood development, December 2014 edition* (pp. 17–24).
- Inclusion Finland KVTL (2015). Incomparable sisterhood. Downloaded October 26th 2015, from http://www.kvtl.fi/contentimages/Julkaisut/KVTL_Verraton_sisaruus.pdf
- Kaulio, P., & Svennevig, H. (2008). Sisaruus – Rakkautta, vihaa ja kateutta (Sisterhood – Love, hate and envy). Helsinki, Finland: Minerva Kustannus.
- Kinanen, M. (2009). *Surusäkki, vertaisryhmätyöskentelyä lasten sururyhmille* (Grief bag – Peer group work in children's grief groups). Helsinki, Finland: Lasten Keskus.
- Kinnunen, T. (2006). Vertaistuki erityislasten vanhempien voimavarana (Peer support as a resource for parents of children with special needs). Professional thesis, University of Jyväskylä, Finland.

- Kolehmainen, T., & Pulkkinen, T. (2009). Sisaruksen kehitysvammaisuus – Vammattomien sisarusten kokemuksia sisaruussuhteesta (Siblings with intellectual disability – Experiences of sisterhood in those who have siblings with intellectual disability). Professional thesis, Jyväskylä University of Applied Sciences, Finland.
- Kulomäki, T. (1984). Pitkäaikaissairas lapsi ja sisarukset (The long-term ill child and siblings). In T. Arajärvi, A. Backman, & P. Siltala (Eds.), *Lapsen psykosomaattikka* (Child psychosomatics). Espoo, Finland: Weilin and Göös.
- McHale, S. M., Updegraff, K. A., & Whiteman, S. D. (2012). Sibling relationships and influences in childhood and adolescence. *Journal of Marriage and Family*, 74, 913–930.
- Meltzer, A., & Kramer, J. (2016). Siblinghood through disability studies perspectives: diversifying discourse and knowledge about siblings with and without disabilities. *Disability and Society*, 31, 17–32.
- Mäenpää, H. (2014). Terveysthuolto, Kela ja RAY sopeutumisvalmennuksen mahdollistajina (Health care, Kela and RAY as an enabler of adaptation training). In H. Streng (Ed.), *Sopeutumisvalmennus: Suomalaisen kuntoutuksen oivallus* (Adaptation training: The insight of Finnish rehabilitation) (pp. 293–337). Helsinki, Finland: Helsinki: Raha-automaattiyhdistys.
- Norio Center (2015). Sisarusten juttuja (Sister stories). Downloaded October 14th 2015, from <http://www.norio-keskus.fi/fi/noriokeskus/sisaruksille>
- Parisaari, U. (2015). Personal communication, Telephone interview, October 14th 2015.
- Petalas, M. A., Hastings, R. P., Nash, S., Hall, L. M., Joannidi, H., & Dowey, A. (2012). Psychological adjustment and sibling relationships in siblings of children with autism spectrum disorders: Environmental stressors and the broad autism phenotype. *Research in Autism Spectrum Disorders*, 6, 546–555.
- Repper, J., Aldridge, B., Gilfoyle, S., Gillard, S., Perkins, R., & Rennison, J. (2013). *Peer support workers: Theory and practice*. London, UK: Centre for Mental health.
- Rubin, K. H., Bukowski, W., & Bowker, J. C. (2015). Children in peer groups. In R. M. Lerner, M. H. Bornstein & T. Leventhal (Eds.), *Handbook of child psychology and developmental science. Volume 4. Ecological settings and processes* (pp. 175–222). New York, NY: Wiley.
- Sarola, M. (2016). Etenevän sairauden vaikutus terveeseen sisarukseen – Terve sisarus tarvitsee tukea ja tietoa (The impact of a progressive disease on healthy siblings – The healthy sibling needs support and information). Masters thesis, Karelia University of Applied Sciences, Finland. (<https://www.theseus.fi/handle/10024/104966>)
- Seligman, M., & Darling, R. B. (2007). *Ordinary families, special children, Third edition*. New York: Guilford Press.
- Trachtenberg, S. W., & Batshaw, M. L. (1997). Caring and coping: The family of a child with disabilities. In M. L. Batshaw (Ed.), *Children with disabilities* (pp. 743–756). Baltimore: Paul H. Brookes.
- Vehmanen, M., & Vesa, L. (2012). Verraton sisaruus (Incomparable siblinghood). *Kehitysvammaisten Tukiliiton Julkaisusarja*, No. 1.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- Western Australian Centre for Health Promotion Research (2010). Peer support. Downloaded December 7th 2015, from <http://mypeer.org.au/planning/what-are-peer-basedprograms/program-types/peer-support/>

Family Organizations and Advocacy Groups

*Barbara Cole, Svein Rokne, Iris Dyck, Margie Frazer,
Susan Fugger, Anne Hagedorn Hamann, Susanne Joensen,
Carrie Mannion, Carsten Munkholm, Sabine Pilgram,
Maria Liisa Punkari and Merete Staureby*

At times our own light goes out and is rekindled by a spark from another person. Each of us has cause to think with deep gratitude of those who have lighted the flame within us.

Albert Schweitzer

Family advocacy groups and other groups safeguarding the interests of individuals with juvenile neuronal ceroid lipofuscinosis (JNCL) play a key role in enabling their members to cope, and thrive in the midst of a severe diagnosis. When parents learn of their child's diagnosis of JNCL, they often are in shock about how this will change their child's life and that of the rest of their family. They ask questions like *How will I care for my child? How did this happen? Why didn't I know how to protect against this? What does this mean?* As the early stages of shock begin to fade into the routine of the "new normal," more questions arise. These big life questions, as well as the more practical questions about how to manage, are most often best answered by other parents who have walked this road with their children. The overarching goal of family organizations and advocacy groups is to help all families facing the disease feel less isolated and to know that others understand their day-to-day worries. They are helped to move from despair to living, from shock to action, and to finding joy again. Parents help each other learn how to travel a new road that was not on the original map for their family. In some countries, the family groups have grown to raise much-needed funds for research and provide educational, social and other support services.

Advocacy groups are organized in many countries to provide special emphasis on the life of the children in school and vocational settings. Here the term "school and vocational setting" is used broadly and includes post-secondary learning, quality of life and learning needs that go beyond the immediate classroom.

This chapter presents advocacy groups related to individuals with JNCL, their philosophies as well as how some advocacy groups provide services to

families and children with JNCL. The chapter includes the parents' associations of Norway, Denmark, Germany, UK, USA and Finland, and a representative of The Royal Blind School in Scotland (Internet addresses can be found at the end of the chapter).

Norsk Spielmeier-Vogt Forening (The Norwegian NCL Family Association)

Norsk Spielmeier-Vogt Forening (Norwegian NCL Family Association), or NSVF, was inaugurated in 1980. In 2018, it has more than 320 members, including most of the 33 children and young adults with JNCL or other NCLs in Norway. Day-to-day business of the Association is managed by a board, elected annually by the annual meeting of the members. In addition, several parent members are engaged in project work and other vital tasks for the Association. Work thus carried out is done *pro bono*.

NSVF is mainly concerned with the «living with» aspect in a life-time perspective, and initiates a number of activities aimed at improving life as it is lived by the families. Every year, there are several events, where parents and other family members meet informally to discuss the various challenges of living with the disease and to provide a place for social interaction among the family members. One of the events is a free holiday during the summer at a holiday resort in the southern part of Norway. Here children and young people with JNCL and their families and service providers can enjoy a week together away from the daily grind. In the autumn, there is a long family week-end – Thursday through Sunday – at a center near Oslo belonging to The Norwegian Association of the Blind and Partially Sighted. Professionals are engaged to facilitate activities such as swimming, boating and fishing in the nearby lake, wall-climbing, ATV driving, horse riding, obstacle routes, and a number of other challenging and enjoyable tasks for the children and young people with JNCL. In addition, there are various entertainments at night; bingo and quiz are always big hits!

The Association initiates and participates regularly in a number of projects. For several years, NSVF has worked with the Oslo University Hospital to provide a national clinical service for children and young people with JNCL. The hospital is now a National Resource Center for NCL, where physicians and parents may obtain guidance and counseling related to NCL diseases. In 2016, NSVF and Oslo University Hospital published *Guide to symptomatic treatment of neuronal ceroid lipofuscinosis*, which can be downloaded in Norwegian and English from <https://www.nsvf.org/Om-NCL-sykdommene/Medisinsk-informasjon-og-oppfølging/Medisinsk-veileder/>.

NSVF's website provides free expert advice to families on topics related to legal rights within the medical, educational, social and psychosocial aspects of living with a child who has JNCL. This service is possible through the Association's collaboration with a law firm which is paid an annual retainer to keep these pages up to date.

NSVF collaborates with Norway's main service institutions in the fields of medicine, education and social work. The Association has regular formal meetings with these organizations, aiming at streamlining services provided to families. This group has developed a set of formal procedures for relaying information about the diagnosis and supporting the family in the first period following this disclosure (see Chapter 24). NSVF has a close cooperation with Statped, a national service for special needs education. The need for early intervention is recognized by this institution, which provides specialized support to schools and other educational institutions throughout Norway.

NCL Danmark (The Danish NCL Family Association)

NCL Danmark (previously Dansk Spielmeyer-Vogt Forening) was founded in 1983. In Denmark there are presently 23 children and young adults diagnosed with NCL. They include one with CLN1, one with CLN2, one with CLN8 and 20 with CLN3 (JNCL). The Association has about 300 members.

The goal of the Danish NCL Association is to provide possibilities for parents to meet and talk with each other about the many issues and challenges they meet in daily life with a child who has NCL. The parents meet three times each year. One weekend is for the general meeting with various courses for the parents. There are nice surroundings and entertainment to promote relaxation and socialisation among parents. One weekend is arranged by the Danish Batten Team (see below) where different problems and issues are discussed. Professor John Østergaard has always participated in this meeting, informing about the latest news in medical research and practice related to NCL. Finally there is one social family weekend with children, support staff, siblings and grandparents. The parents know each other well and there is a very close personal network within the group.

The children and young adults with NCL meet six times each year. This includes one week winter holiday, one week summer camp, one weekend in Legoland (an amusement park in Denmark), the family weekend and two weekends in spring and autumn organized by the Association. These weekends and holidays are very special for the children and young adults with NCL. They know each

other from the very beginning and become best friends. Everyone participates, and there is no differentiation between children, adolescents and adults.

In Denmark, the municipalities and the Government are responsible for providing care services to people with special needs, including people with a rare disease. The social welfare system enables families to choose either to keep their child with JNCL at home during adolescence and adulthood, or live in a sheltered home. In both cases, the young adults will get the help and support they need, and a social pension from the age of 18. At present, most of the young adults are living in their own apartment connected to their parents' home. For their support and care, the municipality provides pedagogical and nursing staff in their home, as well as in schools or activity centers they are attending.

A father's meeting with NCL Danmark

My way to handle mourning is to do things. Just after receiving the diagnosis I wanted to do something that was good for my son. I insisted on all sorts of experiences. We should have a fire and roast sausages. I didn't care whether my son found this okay, I had to feed him with experiences that he could store in his memory. So we did all kinds of things.

The counselor from our Team told about a computer program that could help my son, and in October, 1½ months after the diagnosis, I was on my way to Kalundborg to take part in a course about the program. I knew there would be two other parents present. It was both good and anxiety-provoking.

On the road to Kalundborg I got sick. I had severe stomach pain and had to throw up. I pulled over and thought I'd better turn around, now when I felt so bad. After a serious discussion, I agreed with myself that I was hit by an unknown and irrational fear; that I had to stop thinking and just go on.

In the course room, I sat with 25 other people and looked around. I looked to find the two most sorrowful faces (to find the parents who were present). I didn't find them!

Late afternoon after my first day on the course, I was sitting in the smoking room with my pipe and the door opens. Two women came in and asked whether it is me who is Carsten, the new boy's father. They did not look particularly mournful, but in return they knew exactly how I felt. They were not afraid to make me sad, which meant they could ask me about everything and even help where it was hard for me to explain. It was an indescribable relief.

At this event, a new and life-long learning process started. The very first thing I learned in interactions with parents who were in the same situation,

was that you can have a good life, even if you have a child with JNCL. We eat and drink (a little too much), we laugh and cry, we enjoy and we become wiser. For me, the other parents with children who have NCL are my family. This is true in every way and also includes the parents I have not met yet. In the NCL family there are some you like a lot and some you only meet occasionally. In the family you can talk about everything and you will bear with everything.

The Danish NCL Team

In Denmark there is a unique expert team for NCL, The NCL Team (the Team). The Team mainly exists due to the initiatives of a strong and determined parent association, NCL Denmark. During the 1990s, the Association succeeded in getting the Team started as a temporary project financed by the Government. Today the Team is permanent, financed by the Government.

The Team is cross-disciplinary, which enables it to handle the very complex issues of families with a member who has NCL and to see the problems from different perspectives. There are six members in the Team: one social worker, two special education consultants, one doctor and two parents representing NCL Denmark. The social worker is primarily responsible for social and legal issues in relation to the authorities. This includes the primary contact with social services regarding housing, transport, relief services, pensions and many other relevant tasks. The social worker is also the coordinator of the daily function of the Team. The educational consultants help the families with issues related to school, support staff and after school facilities, and they provide supervision for all professionals working with the children. Each year, the social worker and the two educational consultants organize a three-day course for support staff working with NCL.

The Team's medical doctor is in direct contact with all the children, and provides medical examinations at Skejby Hospital. The doctor is reachable practically around the clock and also makes home visits when needed. If a child with NCL is hospitalized, the doctor always stays in close contact with the local hospital, which in most cases has no experience with NCL. Furthermore, the Team doctor is in regular contact with the local doctors. Most of them have never heard about the disease before. The doctor, as well as the rest of the Team, collects knowledge about all aspects of the NCL diseases.

The parent representatives are elected by NCL Denmark. They are the family experts and know the many complex problems that may arise, because they live in it, live with it and they understand it. They know how it is for the family to be informed about their child's diagnosis, to experience grief, to realize that the child

has problems at school or needs to change school, that there is a need for support in the home or for moving to a residential home, to get a handicap accessible car, and to deal with hundreds and hundreds of meetings with the social services and other public institutions. As Team members, the parent representatives can influence professionals to react and handle the various cases in a more specific and relevant manner, and without losing the human touch. Like the rest of the Team, the parents are bound by professional confidentiality. This is important, because the work of the Team concerns very personal issues. The Team members do their job with great respect for the families they support.

The primary intention behind the Team was to create a support and counseling system for children who had recently been diagnosed and their families. The team has established a set of formal procedures for informing families about the diagnosis (see Chapter 24). It maintains cooperation with the families and with the local educational, health and social authorities that are involved in the daily lives of the children with NCL and their families. The Team is supporting children and adults with NCL and their families throughout life.

NCL-Gruppe Deutschland e.V. (The German NCL Family Association)

In 1977, parents and educators at a school for blind children in Hamburg formed a «community of interests for the advancement and care of children and adolescents with Vogt-Spielmeyer syndrome», the predecessor of NCL-Gruppe Deutschland e.V. In December 1989, NCL-Gruppe Deutschland e.V. was established for children with all forms of NCL diseases and their families throughout Germany. Today, NCL-Gruppe Deutschland e.V. has about 400 members and in 2018 there are eight members with CLN1, 15 with CLN2, 25 with CLN3, one with CLN5 and two with CLN6. Several other families with a child who has a NCL disease get help from the Association without being members.

NCL-Gruppe Deutschland e.V. is a family association and its important functions include providing mutual support, to be there for each other after the diagnosis, to bring the families back on a solid track, and to foster exchange of information and connections between the families who have a child with NCL. The Association provides advice, assistance and support in issues of everyday life as well as special requirements, to the families, their relatives and friends, professionals, teachers and other people who are interested in NCL. This is done through personal conversations and long telephone calls, by email, via the Association's official website and closed group on Facebook, and by distributing information material.

Smaller meetings are organized in four regional chapters, according to the needs and wishes of the members in each region. There is one annual seminar only for mothers and another one only for fathers, with expert speakers on different topics and enough time for the participants to talk and share their thoughts and experiences. For several years, an annual weekend meeting has been organized for siblings of children with NCL, where they can have fun, exchange their feelings and talk about everything in their own peer group.

In contrast to these meetings, the Association's annual conference is for the whole family. The children with NCL and the siblings have their own program, and are taken care of by dedicated caregivers and supporters – with highlights like a visit of a fire brigade with several fire engines. The parents can attend different workshops, talks by scientists, medical and social law professionals, and exchange experiences with other families. They have the opportunity to talk about the needs of their children with experienced doctors from the NCL Counseling Center in Hamburg. The Association works closely with this hospital unit, and families go there regularly with their child with NCL for medical examination and help.

Very important are the two ten-day holiday camps that are organized each year, each with ten children with NCL and lots of dedicated caregivers. The children with NCL can do the same things as other children, teenagers or young adults during this free time, which is different from their ordinary daily life. Over the years, they get to know each other well and they are really looking forward to these camps. The activities may include cinema and concert visits, shopping, swimming, barbecue, bonfire, listening to and making music, or visit to a leisure park, including a roller coaster ride.

Through its participation in the present ERASMUS+ project on education and JNCL (the JNCL and Education Project), NCL-Gruppe Deutschland e.V. seeks to improve education and communication for children with JNCL and give caregivers and teachers better insight into the special needs of the children with JNCL. Germany is a federation with 16 states, each with a different school system, and it is a hope that the ERASMUS+ project will help provide the best education for children with NCL in all the states. Moreover, to achieve the best outcome of education, an early diagnosis of NCL is essential. Through several activities, the Association endeavors to increase knowledge and awareness about the disease and thus the probability of early detection and diagnosis and thereby to provide the best possible support and therapy in daily life and school. Information material is sent to hospitals, pharmacies, therapists, students and doctors. NCL-Gruppe Deutschland e.V. tries to inform journalists, has information stands on fairs, in banks and town halls, and organizes events in several German cities on the Rare Disease day at the end of February. The Association also tries to get a foot into the symposia for doctors, to provide information that might raise their awareness not

only of the challenges imposed by NCL diseases, but the potential for a rich life despite the consequences of the disease.

Apart from providing public education services to diverse audiences, the Association is increasingly involved in NCL research, international networking, and legislation. Although the Association recognizes that coping with NCL diseases is an ongoing reality, it also seeks to draw attention to how people with these conditions, together with their families, can participate in satisfying life experiences.

Batten Disease Family Association (BDFA)

The BDFA is the only UK National Charity that supports families who have a child, adolescent or adult with JNCL. Although the team is small, the BDFA supports families across the UK, providing holistic support to ensure that children and young people with JNCL receive the care and support services that they will need in different phases of the disease.

The role of the support and advocacy team is to interact with families from the point of diagnosis and throughout life. This can be through visits to the family home, via emails or phone calls and accompanying them to large multi-disciplinary team meetings. Often families are not given much of an explanation of the disease from the hospital and are often left with feelings of shock and loneliness. The BDFA provides more detailed information to families and can give anonymized examples of other family situations that may be similar to their own to provide answers to difficult questions and to help families feel that they are not alone.

The BDFA provides holistic, person-centered practice for families and individuals. Not only does the BDFA look at the needs of the child but also the needs of the wider family around them and the needs of the professionals working with them. The BDFA aims to empower and build resilient families to be able to share their views at meetings which sometimes have over fifteen professionals attending, and to help build a solid support network around them. Families often have medical questions that sometimes their local team struggles to answer. The clinical nurse specialist of the BDFA is able to talk to families if their local team cannot provide sufficient answers to their questions.

It is important for families to feel empowered when making decisions about their child's care as well as being able to make others aware of their child's needs. The BDFA offers all families support by providing an information packet called a *family folder*. This folder gives a brief medical description of the different symptoms of the disease and information on education, social care, research and genetics. The feedback from families about the folder has been positive. Many take the folder

to meetings with professionals who know nothing about JNCL and parents take it with them if they unexpectedly have to go to hospital in an emergency, so that the staff there can quickly learn about the symptoms and management of the disease.

The BDEFA provides emotional support to the family and the extended family through a free phone helpline. This helpline is also used by professionals who work with young people with JNCL for emotional support and for advice, signposting and guidance. School visits form a significant part of the support and advocacy program. Often, the children with JNCL are in mainstream school when they are diagnosed. The BDEFA works with these schools to provide specialist training on JNCL and to provide practical strategies that the schools can use. This may include behavioral strategies, information on JNCL and visual impairment, and how to work with young people with dementia.

Social isolation is often an issue for families with a child or adolescent who has JNCL. Often families feel alone as they do not know of any family in their area with a child with the same disease. Social media play a huge part in reducing that isolation for the families, and the BDEFA often puts families in contact with each other or adds them to various secure discussion groups on websites for parents of children with JNCL.

The BDEFA holds an annual family conference which is free for families to attend. Professionals from across the UK also attend the conference to speak with families about education, research, social care, and health care. Families value this time to be able to get together with professionals and with families they may have only met through social media. The BDEFA recognizes that for most families this is not easy. Parents with children who were recently diagnosed will see young people who are much further in the disease progression than their own child. Families with older children will see children who are in the initial phases of the disease, and bereaved families may be attending the conference for the first time without their family member with JNCL. However, despite being at different stages, they value the time that they spend together, sharing their experiences and gaining knowledge from others who have been in the same situation. For bereaved families, there is space for reflection in the remembrance room.

At the conference, the BDEFA provides a complete program of care and activities for children and young people, enabling them to interact with one another, for siblings to gain support, and for parents and caregivers to access the professionals in the knowledge that their children and young people have professional support.

Another function of the BDEFA is to support families through decisions about their young person's care needs and where they will be living once they reach 18 years. Many families choose for their young person to remain with them at home, although some young people go to live at Heather House, a care home which has provided care for many young people with JNCL over the past few years. The

BDFEA also supports families to find a counselor who knows about JNCL, as many of the local counselors are not familiar with the disease and families can find this very unhelpful.

Collaborative working is the core to the way in which the BDFEA delivers its services to children and families. The Association does not believe that it can or should try to support families on its own; the BDFEA will continue to advocate on behalf of the families, working with other disability organizations to improve services and care for children and young people with all types of NCL diseases across the UK, because it is only together that stakeholders can make a difference.

Notes from an educator in Scotland

Children and young people with JNCL often build up what to parents is an alarming amount of documentation about the provision of education and therapy for their child. Pupils in Scotland, depending on their local authority, will have a Plan which should encompass all aspects pertinent to their situation (see also Chapter 11). In a School, such as The Royal Blind School, there will be review meetings regarding the education, health and care of the pupil. These reviews take place at least once a year and more frequently as required, for example in preparation for transitions.

For parents these reviews may be daunting owing, in part, to the number of team members present, which can exceed ten. Around the table may be a community pediatrician, an educational psychologist, local authority professionals in charge of placements for pupils with additional support needs, a social worker, a physiotherapist, a speech and language therapist, an occupational therapist, a habilitation specialist, a class or form teacher, a deputy head teacher and a principal teacher. At a time of transition, there will also be professionals from the service the pupil will be attending. At one such review, twenty professionals attended. The parent found this overwhelming and did not attend the next reviews, even after reassurance that the numbers would be fewer. This parent had been encouraged by the school to liaise with the BDFEA but could not be persuaded. The potential for a difficult transition process for the adolescent increases when the family and the team do not share goals or strategies for attaining them.

The experience of other parents was very different because they engaged with the BDFEA once they had a diagnosis for their child and before the child came to the school. The role of the BDFEA cannot be overestimated. The BDFEA worked with the parents to communicate with the school and placement authority and the professionals responsible for implementing placement. For any parent the BDFEA is also a link to meeting other parents and to attend weekends where the

whole family is welcome and can share their concerns whether as a parent or a sibling. The school's link with the BDEA ensures that the most effective practice in education for children with JNCL is shared and any new ideas or innovative practice can be shared. This kind of professional dialogue is vital to maintaining the highest standards of learning for all children and young people with JNCL.

The Batten Disease Support and Research Association (BDSRA)

Since 1987, the BDSRA has been a guiding force in advocacy for all forms of NCL. Funding research, supporting parents and developing programs that go to the heart of families' needs have been keys to the organization's success.

BDSRA reaches families across the US and world in a variety of ways. Social media, through Facebook, Twitter and Instagram provide platforms to report news, respond to questions and provide educational content to families and loved ones. At this writing, there are 900 family members and caregivers from different parts of the world on the BDSRA private page, monitored by staff members.

Families also connect in person each year at the annual three-day family conference, which includes programming for parents, extended family members, young siblings and researchers. Workshops, celebrations and remembrance are all a part of this event. Because so many are in attendance, researchers are able to engage with families to ask questions, complete natural history information about the disease, and collect blood and saliva samples. These data are keys to scientific progress.

In addition to ongoing clinical referrals for parents, school consultations for teachers, and clinical trial patient support, the BDSRA is keenly involved in research development. Parents and supporters have funded research projects for all forms of the disease, leading to some of the human clinical trials moving forward now. Each of these families raised money for research they knew would never help their own child, but that might benefit children with NCL diseases of the future.

Each year the BDSRA performs a basic and translational research merit review process, requesting proposals from around the world. Each proposal is reviewed by experts in lysosomal disease, pharmacy, genetics, pediatric neurology and other fields. Those with the highest scores are funded by BDSRA and family and other foundation partners. Family foundations such as Noah's Hope/Hope for Bridget, Our Promise to Nicholas, Beyond Batten Disease Foundation, Drew's Hope and Taylor's Tale among others fund research for the type of NCL disease their children have had.

BDSRA and other family foundations provide consultation to researchers, industry, and regulatory agencies to help them understand the voice of individual persons with JNCL in drug development. The Association does this by organizing focus groups, recruiting families to speak at public hearings, and providing letters of support for legislative efforts important to the families, such as healthcare coverage, newborn screening and improved regulatory processes to further rare disease scientific exploration.

Support, research and advocacy are the pillars of BDSRA, driven by Batten family social action, funding and hope for a brighter future.

The Finnish NCL Family Association

The Finnish NCL Family Association was registered in 2007 but has worked actively since the 1980s as a group of families with children who have JNCL. The main aim of the Association is to share information about JNCL and provide peer support to the families. The Association has about 100 members, including 35 families with a member who is diagnosed with JNCL (CLN3), two with late infantile neuronal ceroid lipofuscinosis, LINCL (CLN2 and CLN5), 20 external members and two company members. There is a separate group for infantile neuronal ceroid lipofuscinosis, INCL (CLN1), with around 40 families with a child who has this diagnosis.

The Finnish NCL Family Association organizes four to six events each year, including one or two events for families, one event for parents only, and weekends for mothers and fathers. In those events, there are usually some lectures by professionals and official annual meetings, but the main aim is to be together and have valuable peer support. The Association shares information via their webpage, email and events. It organizes a Facebook group, which is also a good way to share information and provide peer support.

The Finnish NCL Family Association has a history of strong relations with the Finnish Federation of the Visually Impaired (FFVI), which provided an NCL specialist for families until 2015. In 2017, the Association started working with the Norio Centre of rare and genetic disorders, including funding for an NCL specialist in one year. The continuation of this position depends on future funding. The Association is not getting regular funding and all activities and events are organized and managed by members of the Association. The lack of publicity and keeping a low profile are hindering the recruitment of sponsors, but raising the profile may have a negative effect for some families.

Links to the associations

Batten Disease Family Association	http://www.bdfa-uk.org.uk/
Batten Disease Support and Research Association	https://bdsra.org
Bildungszentrum für Blinde und Sehbehinderte in Hamburg	https://bzbs.hamburg.de
NCL Danmark	http://dsvf.dk
Norsk Spielmeyer-Vogt Forening	https://www.nsvf.org/
Oppimis-ja ohjauskeskus Valteri, Onerva	http://www.valteri.fi
Royal Blind School	https://www.royalblind.org/education
Statped	http://www.statped.no/
The Finnish JNCL Family Association	https://www.jncl.fi
The German NCL Family Association	http://www.ncl-deutschland.de/
Vision Centre Refsnæs	http://synref.dk/
WESC Foundation	https://www.wescfoundation.ac.uk

Behavioral and Emotional Reactions to a Difficult Life Situation

*Stephen von Tetzchner, Jochen Lippe Holstein
and Per Kristian Haugen*

Juvenile neuronal ceroid lipofuscinosis (JNCL) is a serious and complex disease. Children and young people with JNCL experience a sequence of challenges, each of which may have a pervasive influence on their lives. First comes the onset of visual problems and gradual complete loss of vision, then academic problems in school, followed by motor and language problems. These impairments often lead to reduced participation in activities with peers and friends. Over time, children and young people with JNCL will realize that their own development is following a different course from that of others. Their achievements are falling behind, resulting in an increasing gap between their abilities and those of their peers. They may experience frustration and feelings of loss and insecurity related to what is happening. It is important that such feelings and the reactions of the children and young people are met with special attention and understanding. The present chapter discusses these reactions and how children and young people with JNCL can be helped to cope with their life situation in the best possible way.

Emotional and behavioral reactions and JNCL

Children and young people with JNCL will react in different ways to their difficult life situation, both emotionally and behaviorally, and with internalizing or externalizing reactions. Some of them withdraw from some or many situations, rejecting suggestions for activities that they might otherwise find very attractive. Others react strongly (i.e., overreact) to the slightest demands, possibly because of fear of failure or being an outsider, and may show aggression towards other people or objects. Sometimes such behaviors occur without an apparent cause. There are however individuals who show resilience by adapting and coping in spite of many challenges (Goldstein & Brooks, 2013; Rutter, 2013), and expressions of resilience may also be found among children, adolescents and adults with JNCL.

Although there are few studies addressing the emotional and behavioral reactions in individuals with JNCL, those studies are consistent in the finding that there is a higher level of emotional and behavioral problems in this group than in peers with typical development (Adams et al., 2006, 2013; Marshall et al., 2005; Santavuori, Linnankivi, Jaeken, Vanhanen, Telakivi, & Heiskala, 1993). Adams and associates (2006) asked parents of 25 individuals with JNCL, aged 6 to 18 years (average 12.3 years), to complete *Child Behavior Checklist* (CBCL, Achenbach & Rescorla, 2001) and *Scales of Independent Behavior* (SIB, Bruininks, Woodcock, Weatherman, & Hill, 1996). On the CBCL, the highest scores (indicating more severe problems) were on the subscales «Thought problems», «Social problems», «Somatic complaints» and «Aggressive behavior». On the aggression subscale, three participants were given scores in the clinical range and seven in borderline clinical range. The average scores for «Anxious/depressed» and «Withdrawn/depressed» were close to the general population mean of the same age. On these two subscales, one individual had a score in the clinical range and five others had scores in the borderline clinical range. Among the 20 most frequent items that were «somewhat true» or «often true» were «Can't get his/her mind off certain thoughts, obsessions», «Demands a lot of attention», «Argues a lot», «Talks too much», «Temper tantrums or hot temper», «Clings to adults or too dependent», «Sudden changes in mood or feelings», and «Impulsive or acts without thinking». On the SIB, one was given a score in the serious range and nine in the moderately serious range on the «Asocial» subscale (two items: a) Socially offensive behavior and b) Uncooperative behavior). Two individuals scored in the moderately serious range on the «Externalizing» subscale (three items: a) Hurtful to others, b) Destruction of properties and c) Disruptive behavior). None scored in the moderately or more serious range of internalizing behavior (three items: a) Hurtful to self, b) Unusual repetitive behaviors, and c) Withdrawn or inattentive behavior). The scores on the two checklists were a little above the average, indicating somewhat more problems than in the general population. However, it is important to emphasize that checklists do not represent a diagnosis. They were completed by the parents and the items on CBCL were not adapted to individuals with severe visual impairment.

In a Finnish study with 27 slightly older children and adolescents (mean age 15 years, range 9–19), the most prominent behavioral features were problems of social interaction and aggressive behavior (Bäckman, Santavuori, Åberg, & Aronen, 2006). Similar to the study of Adams and associates (2006), symptoms of depression were reported to be rare. A study with 42 children and adolescents (mean age 10.2, range 5–15 years) found that restlessness, aggressive behavior and fear were common features (Santavuori et al., 1993). Difficulties with sleep were especially addressed by Santavuori and found to be a common problem. Depression was reported for four of the 42 individuals but Santavuori considered

unrest, aggressive outbursts, anxiety and self-injury as symptoms of depression. Combining several studies comprising 70 children, Adams and associates (2013) found a small increase in raw scores on the subscales «Social problems» and «Aggressive behavior» on the CBCL from early to later childhood, and a decline in scores on these two subscales from late adolescence into adulthood. However, these averages hide considerable variation.

In the present study (Appendix A), parents were asked about various emotional and behavioral problems. About 75 percent of the participants with JNCL had shown some kind of emotional symptom (anxiety, depression, fear, etc.). These reactions tended to appear for the first time in childhood, but some appeared first in adolescence and early adulthood (Figure 27.1). Common emotional reactions were anxiety, depression, passivity and excessive sadness. As one could expect, there was sadness due to changing social settings, loss of interaction with peers and loss of friends – but still relatively high contentment despite the disease.

It is understandable that many children and young people with JNCL react with anxiety, depression, and fear. Anxiety is a typical reaction to the unknown. For example, individuals with autism spectrum disorder have difficulties understanding other people and why they act as they do, and anxiety is very common in this group (Kerns & Kendall, 2014). Depression may reflect a state of lack of power, of having little influence on decisions about one's own life, and low mood and passivity are frequent reactions in this group. Anger may be an expression of fighting back and not accepting what is happening. Depression can also express itself in agitated and disruptive behavior (American Psychiatric Association,

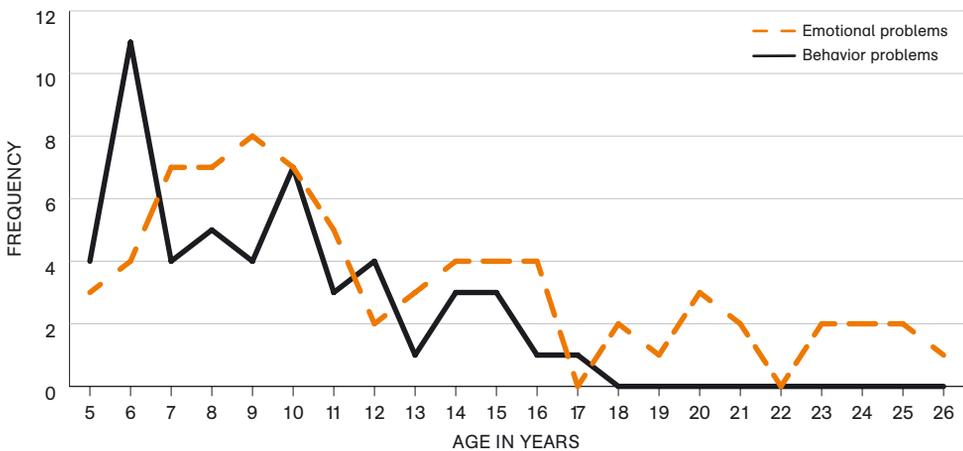


Figure 27.1 Frequency of subjects at each age when emotional reactions (anxiety, depression, fear etc.) and behavior reactions were noticed for the first time

2013), and Santavuori and associates (1993) point to unrest, aggressive outbursts and self-injury as symptoms of depression.

Behavioral reactions were observed less frequently than emotional reactions, in about 56 percent of the participants, but the distribution of first observation followed a similar pattern (Figure 27.1). The most common behavioral reactions were verbal and physical aggression, while spitting, swearing, object destruction and self-injury were less common (Figure 27.2).

The results from the present study with a relatively large sample of individuals with JNCL demonstrate that the disease often leads to emotional and behaviour reactions but also the significant variation in such reactions within this group.

Many research studies measure negative reactions to life events without providing parallel measures obtained from rating scales for positive responses. A positive attitude to life is indicated or assumed by a lack of emotional and behavioral problems. However, everyday life is usually a balance between positive and negative elements, and this situation is also true for individuals with visual impairment and cognitive challenges. Only focusing on negative elements will not sufficiently represent the overall situation of the child or young person with JNCL. This biased profile is well illustrated in the quotation below.

Our daughter was never aggressive or ungrateful. She accepted her disease. She was always friendly, said «please», «thank you» and «no thank you» and is very well liked at school, in her residence and in her leisure time activities. She likes many things. Eating is most important for her; she loves

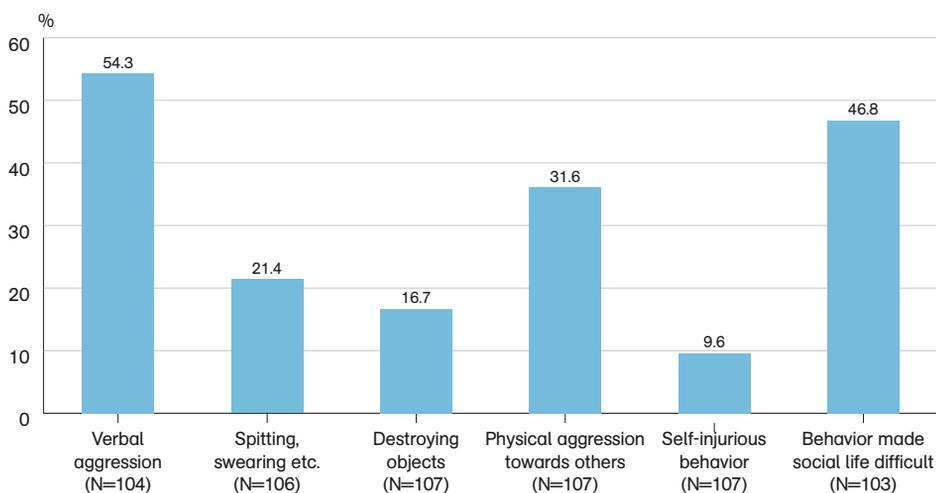


Figure 27.2 Percentage of participants who had shown different forms of behavior problems

her gherkins (58 kg weight, 162 cm height). She loves to watch her Harry Potter and Walt Disney DVDs, and she knows everything by heart, speaks along and laughs about the same passages. She is a cheerful and kind-hearted person. Her laughing comes from her heart. She is grateful for every little bit, and answers this with «thank you», «no, thank you» or «please».

In the present study, parents were asked about the frequency of four major moods in their child at different age levels. Figure 27.3 shows that it was a positive and enthusiastic mood that characterized the individuals with JNCL in almost all ages, although the scores of troubled, anxious and irritated and annoyed moods increased with age.

Children and young people with JNCL have more challenges than others. The small number of earlier studies and the present study (Appendix A) have found that many in this group react to their difficult life situation in different ways, emotionally and behaviorally. The large variation suggests that the brain disease in itself is not responsible for all the reactions and so it is necessary to investigate the influence of other factors, such as the quality of the physical environment and the responsiveness of people in the environment. These will influence the well-being and the emotional and behavioral reactions of children and young people with JNCL, as well as how these reactions are met by people in the environment. This complex interaction can best be described as a transactional process (Sameroff, 2010). In the present survey, parents reported one common compensatory strategy they favored was to try to reduce stress, as illustrated in this quotation.

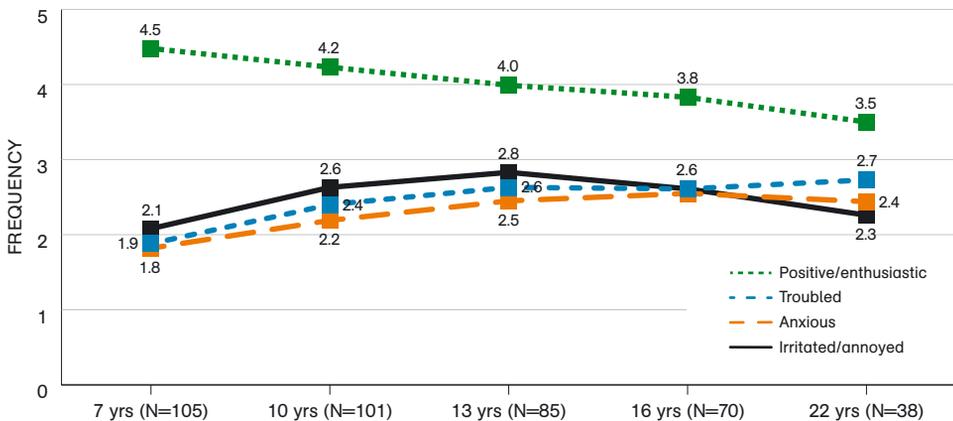


Figure 27.3 Frequency of different moods across age

Scale used: 1=Never, 2=Seldom, 3=Sometimes, 4=Often, 5=Very often

The environment was extremely shielded, all wishes were fulfilled. Therefore the behavioral disorders had no or only sometimes a negative impact.

We avoided situations which meant a lot of stress to her. So we stayed at home, in the garden, cared for pets, long walks (later in the wheelchair). With the dog in the woods and along the lake. We did not make any plans in advance but we did the things that she wanted to do, like handicrafts, meeting people of her age, or caring for animals.

This finding emphasizes the importance of three key needs: assessing the various declines, ascertaining how to influence the everyday functioning of children and adolescents with JNCL, and establishing how support may prevent or reduce secondary effects of the various features of the disease.

Vision loss, dementia and behavioral and emotional reactions

Blindness is a core symptom of JNCL which leads to uncertainty and a lack of situational overview. The consequences may be further aggravated by another core symptom, cognitive decline, which makes it difficult to explain to the individual in meaningful ways what is happening to them and how they can cope. People and objects alike lose their habitual appearance and orientation, rendering mobility more difficult (see Chapter 16). Information about what is going on in the wider surroundings must now be obtained through the use of hearing. People typically rely on visual cues to know who is in the environment, what they are doing, and what activities might be possible. These factors influence their choice of topic and patterns of social interaction. Without the visual cues, many participation opportunities go unnoticed, with severe consequences to the child's play, collaboration and communication, and school performance.

Frustration and misunderstanding

It is a general finding that frustration may lead to aggression, hopelessness and despair (Breuer & Elson, 2017). The assumption that the reported behavioral reactions were a result of a difficult life situation was supported by comments from parents in the present project (Appendix A), who pointed to frustration and misunderstanding as common causes of anger and aggression.

When reality turns out to be different from what she had imagined.

He is an amazingly well-balanced young man who only reacts in an aggressive way when he feels misunderstood. This is often due to his massive loss of speech. He still is full of beans. It seems as if he wants to fill every minute with important stuff without any breaks (whether during the day or the night).

Transition situations with potential loss of control were also mentioned as difficult situations which might lead to emotional or behavioral reactions.

A young person with JNCL may show anger and desperation due to writing problems, and, for example, throw the braille machine off the table or insult fellow students. In such situations, a soothing distraction can be that together, the teacher and student leave the situation, take a break or change to tasks over which the student feels more mastery. Singing a familiar song or recounting a fondly remembered event were often mentioned as useful strategies in the present study, which may change both the general atmosphere and the student's mood.

Anxiety and depression

The life situation of children and young people with JNCL also implies a vulnerability for emotional problems. Losing vision may be experienced as dramatic: «Fear is a logical symptom, because loss of vision elicits both anxiety and insecurity and children experience the environment as dangerous» (Santavuori et al., 1993, p. 247). The results in the present project (Appendix A) indicate almost the same prevalence of behavior problems among children and adolescents with JNCL as with congenital blindness, around 50 percent. Whereas behavior problems among children with congenital blindness are related to missing or delayed social skills (Alon, Ophir, Cohen, & Tirosh, 2010; Runjic, Prcic, & Alimovic, 2015) early behavioral difficulties in children with JNCL (e.g., disruption or verbal aggression) are reactions to frustration. In the present study, few participants with JNCL showed any physical aggression towards others and very few children showed self-injury.

In adult dementia, emotional and behavioral disorders are related to how the individual person experiences changes in competence that accompany cognitive decline (Cerejeira, Lagarto, & Mukaetova-Ladinska, 2012). The disorders are not a direct result of the changes in the brain; instead anxiety and depression reflect reactions to changes in personal competence in routing occupations of life. Depression is a frequently cited symptom throughout the course of dementia and relates to both psychological and biological factors (Lyketsos & Olin, 2002). Key factors contributing to depression include failure to master new ways of accomplishing familiar tasks and inability to adapt to changes (Enache et al., 2011).

In the earlier stages of dementia, people are aware of their difficulties remembering everyday things and decreasing ability to complete the basic activities of daily life. This awareness phase is accompanied by growing reactions of frustration. However, with increasing severity of the disease, they may become less aware of the changes they experience and the source of their frustration possibly shifts from an internal to an external focus (see Chapter 5).

Another population that experiences anxiety, depression, and behavioral disorders includes high-functioning children with autism spectrum disorder (Joshi et al., 2010). Within this population, these disorders may be related to confusion originating in the children's problems with mind understanding and general cognitive problems. By contrast, the visual impairment of children and young people with JNCL leads to less control and more uncertainty, and the cognitive decline makes it difficult to find new ways of doing things and coping with issues. As in adult dementia and autism spectrum disorder, reactions may be observed as forms of anger and aggression, as well as passivity and withdrawal. This implies a need to ascertain the factors underlying behavioral reactions.

In the present study (Appendix A), parents were not asked to distinguish between anxiety and depression because the distinction may not be discernable to a lay person. However, the fact remains that anxiety and depression are different problems, despite sharing symptoms, and frequently presenting as comorbid states: while children with anxiety are wary of potential threats, children with depression ruminate on hopelessness and loss (Hankin, 2012; Hankin, Gibb, Abela, & Flory, 2010). Children and adolescents who are depressed show less pleasure, interest and concentration than they normally would, and easily become tired. They sleep poorly and have little appetite, low self-esteem and little confidence. In the case of JNCL, having little confidence may be both an expected reaction to loss of functional competence in everyday tasks, and a symptom of a resulting depression. Depression can become deeper when it leads to social isolation and having fewer friends. The inability of adolescents, as a group, to solve the problems they meet may contribute to typical symptoms, such as stress, a poor self-image and dysphoria (Hankin, Young, Gallop, & Garber, 2018); however, atypical symptoms include restlessness, anger, irritation, weight gain and increased sleep (Ballard, Day, Sharp, Wing, & Sorensen, 2008). A similar atypical pattern of symptoms may be observed in young people with JNCL.

She oftentimes argues and just has her way of thinking about things and is not receptive to other ways of thinking about it at this point. So there are times where she sits in the breakfast room and has to be moved to a different table, you know. We have put her on a table with girls, eighth grade girls sometimes, in breakfasting and that seems to help because she doesn't seem as confrontational with those kids.

Later, as the disease progresses, adults with dementia tend to become increasingly passive, not taking initiative and are seemingly not interested in participating in activities that they previously liked to engage in. Passivity may also function as a way of ensuring that nothing bad happens, even if it also means that nothing good happens. The person with dementia keeps control by being passive. Some become more impatient and irritable. It can be threatening and very frightening to experience not understanding or having control of one's own situation. Some people experience panic-like anxiety, typically in situations where they notice that they come out short on tasks that they previously have managed without difficulties (Badrakalimuthu & Tarbuck, 2012; Goyal, Engedal, & Eriksen, 2019). In young people with JNCL, anger over legs that can no longer be used for safe walking can lead to vehement pounding on the thighs. Word-finding or articulation problems can lead to aggressive rage or desperate crying – or to a total lack of communication. Individuals with JNCL may have to adapt to a new life situation due to further physical and mental changes, so they can feel safe and comfortable again. But then there is another change in the situation: young people with JNCL are repeatedly being confronted with something happening to them which they cannot control.

It was always important to her to be independent. She always got very angry when she was not able any more to manage things on her own. It was difficult for her to accept help.

The result may be insecurity, anger, fear and grief, all of which could be accompanied by externalizing reactions.

Children and adolescents spend a lot of time at school, where their problems often become more apparent; and where their depression is typically aggravated by an awareness of increasing sense of social isolation and having fewer friends than they had previously. When adolescents are unable to solve the problems they face, they experience even more stress, low self-esteem and depression (Roberts, 2015). In severe adult dementia, there may be behavioral changes in the form of suspicion, delusions, socially inappropriate behavior, impulsivity or emotionally indifference (Steinberg et al., 2008). Such reactions may sometimes be due to misinterpretation or misunderstanding due to the cognitive impairment and a reduced overall grasp of what is going on. Similar reactions are recognizable in adolescents and young adults with JNCL.

The special behavioral issues mostly occurred in early adolescence. They were not permanent and did not affect life in a special way.

Parents and teachers may experience situations and behaviors that are not easy for anyone involved to handle. The children and adolescents with JNCL can suddenly behave aggressively towards others or against themselves, as well as become extremely withdrawn, sad or closed. The reason for the behavioral reactions is not always apparent.

Attachment

Attachment represents another perspective to understanding the emotional and behavioral reactions of children and young people with JNCL. They may sense not only their own lack of control but also their parents' lack of control: the parents are no longer omnipotent. The attachment system may be relevant for their reactions. According to Bowlby (1982), attachment is an innate behavioral system for ensuring protection. He defines attachment behavior as «any behavior that results in a person attaining or maintaining proximity to some other clearly identified individual who is conceived as better able to cope with the world» (Bowlby, 1982, p. 669). Attachment behavior may be activated by unfamiliar people, places and routines, pain, fear and stress. Illness and fatigue increase the need for protection and will heighten children's need for an attachment figure – a parent or another close person (Bowlby, 1969, 1982). Events occur in which children or young people with JNCL feel a need for protection but feel that help is not available and that there is no secure person to turn to. Experiences with events such as these may reflect that the persons with JNCL do not have the psychological protection that attachment implies. One parent described this in her daughter:

When she was six years old she started to develop separation anxiety.

Central to separation anxiety is the fear of being separated from attachment figures. Children may cling to caregivers and react with angry outbursts, kicking, screaming and crying when someone tries to separate them. Children may also experience physical symptoms when leaving for kindergarten or school, or in other ways refuse to go to school. Adolescents can be anxious to be left at home alone.

Anger may also be a reaction to an experienced loss of protection, to the unavailability of protection from an attachment figure. The anger has two functions: first, to empower the child to overcome the obstacles on the way to reunion with the attachment figure and security, and second, to try to ensure that the attachment figure will be available in the future. This type of anger does not destroy the emotional bond between child and attachment figure, but instead strengthens it. Since fear and anger are activated under similar conditions, they often occur simultaneously, and an increase in the intensity of one emotion can

at times strengthen the other (von Tetzchner, 2019). It has been suggested that challenging behavior in children and adults with intellectual disability and autism spectrum disorders sometimes reflects attachment reactions (Janssen, Schuengel & Stolz, 2002; Perry & Flood, 2016; von Tetzchner, 2004).

Exploration is necessary for learning about the world, and exploration is activated by unfamiliar or complex objects and locations. The usual driving forces for exploration (i.e., novelty, unfamiliarity) are identical to the forces driving children to seek an attachment figure (the unknown and uncertainty). Exploration and attachment seeking are thus activated under almost the same conditions, but attachment behavior is elicited mostly by fear and insecurity, while exploration takes place when children feel relatively secure. When the attachment figure moves further away from the child, attachment behavior is activated, while exploratory behavior is more easily activated when the attachment figure approaches, for example in situations involving fascinating objects such as toys or animals. By means of attachment, familiar adults become a secure base for controlled exploration of objects and locations that otherwise can elicit insecurity or fear, a base the child alternately leaves and returns to (Ainsworth, 1963). Thus, attachment and exploration are to some extent complementary ways of meeting new situations, meaning that exploratory behavior is inhibited in situations in which children show attachment behavior.

The attachment and exploratory systems emphasize the need to develop security-giving structures that allow children and young people with JNCL access to social and societal participation. Self-directed exploration may be independent or interdependent (see Chapter 16), and supporting exploration may include activity-based education and adapted tasks that allow students to learn through their own discoveries. Exploration may also imply combining established knowledge with new knowledge. For students with JNCL, exploration may function as an activation of prior knowledge and thereby refresh their memories of earlier experiences. This may range from identifying simple tactual or auditory sensations to investigating whole or parts of complex events, which the teacher may help to integrate (von Tetzchner, Fosse, & Elmerskog, 2013). When the student with JNCL has been guided to explore the whole school and has become better acquainted with peers and staff, the school environment will be experienced as secure, which in turn may prevent emotional and behavioral reactions in children and adolescents with JNCL.

Reactions to loss of friends

As the disease progresses, interaction with peers tends to be reduced (see Chapter 22). Friends are central to young people's lives, and the breakup of a close friendship can be a difficult experience and lead to depression, guilt and anger

(Rubin, Bowker, McDonald, & Menzer, 2013). Furthermore, depression is often accompanied by other impairments, including emotion regulation disorders. Psychological problems such as anxiety, depression, and poor self-esteem are all related to experienced loneliness. Since emotion regulation has a social basis, regulation disorders can have a serious impact on children's social development. Some years into school age, children begin to compare their skills, attitudes, possessions and families with those of their peers; however, low self-esteem based on such comparisons can lead to depression (Harter, 1987). This in turn may lead to cascading effects of decreased interest and motivation to participate in peer activities, lower activity levels, and little support and attention from the environment. This emphasizes the need for structure, control and positive challenges within a recognizable and familiar framework.

When social contacts are lost and new ones are difficult to establish and build, changing school may sometimes help.

His emotional and behavioral problems got better when he changed from regular school to the LBZB (school for students with learning difficulties).

If loss of friends becomes absolute, this may be experienced as dramatic and traumatic, and the result may be comparable to post-traumatic stress disorder (PTSD), a reaction to a stressful or traumatic event a child has experienced or learned about. Children with PTSD can have intrusive memories and recurrent disturbing dreams with fear. The distress is persistent and intense, and the child finds it difficult to calm down. They can feel a great deal of shame, guilt, fear and confusion, and express few positive emotions. They tend to be irritable and angry, are easily startled, and may have difficulties with sleeping and concentrating. They also participate less in play and other positive activities. The symptoms of PTSD involve unregulated emotions and can be reminiscent of behavioral problems. Because some of the reactions are the same, PTSD can be confounded with depression.

Concluding comments

Studies show that the prevalence of emotional and behavioral problems is higher in individuals with JNCL than in the general population. These reactions are related to their declines and the consequences these declines may have for coping in everyday activities and participation in social life. Many factors contribute to these reactions which may be understood from different perspectives, including insecurity, confusion, loss of personal power, control, self-efficacy, and loss of

experience of protection. However, the studies also show there is considerable variation in the scope and severity of these reactions; and that support and adaptation of the physical and social environment may prevent or reduce emotional and behavioral reactions.

It is therefore of critical importance to seek to implement preventive measures. In the present study, behavioral reactions were reduced for some students with JNCL after adaptations were made in the environment to meet the student's social and learning needs; for instance, efforts to compensate for the loss of vision helped the situation, suggesting that the reactions were related to the loss of mastery. Both educational strategies and the social environment of the school are important in this connection, not only in early childhood but throughout school and after the individual has left school. For example, during adolescence, mood fluctuations are quite common in the general population (Laugesen, Dugas, & Bukowski, 2003; Zeman, Cassano, Perry-Parrish, & Stegall, 2006). Thus, it may also be a period where young people with JNCL need special consideration.

An important aim is to provide measures that may help persons with JNCL create meaning and coherence in their lives. One approach is to create a biography with important contacts, themes and experiences, which can enable the person with JNCL, family and staff to remember and use information of special significance to the person with JNCL, and thereby constitute a foundation for security, best possible life flow and meaning-in-activity (see Chapters 11 and 23). Particularly when memory problems are emerging, events that are significant for the individual can be used to call forth pleasant memories and access to familiar structures of action. The following quotation illustrates both the challenges contributing to emotional and behavioral reactions, and a solution to cope with the challenges.

In retrospect the psychological stress during the process of blindness (from 7 to 14 years until adolescence) reached its peak when she had to experience consciously that she was not able to keep up with the others any more although she still wanted to be part of them. At first she became disappointed, sad and angry, then she felt resignation and a kind of acceptance in the end. At this stage she wrote a diary and invented stories which focused on herself and the desperate plight because of the increasing blindness. Writing seemed to help her cope with the disease. That is why she did not suffer from very big psychological problems. She even regained her zest for life. At school and in the sheltered workshop she was known for her cutting remarks or her derisive laughter.

References

- Achenbach, T. M., & Rescorla, L. A. (2001). *Manual for the ASEBA School-Age Forms and Profiles*. Burlington, VT: University of Vermont, Research Center for Children, Youth, and Families.
- Adams, H. R., Mink, J. W., & University of Rochester Batten Center Study Group (2013). Neurobehavioral features and natural history of juvenile neuronal ceroid lipofuscinosis (Batten disease). *Journal of Child Neurology*, *28*, 1128–1136.
- Adams, H., de Blicke, E. A., Mink, J. W., Marshall, F. J., Kwon, J., Dure, L., Rothberg, P. G., Ramirez-Montealegre, D., & Pearce, D. A. (2006). Standardized assessment of behavior and adaptive living skills in juvenile neuronal ceroid lipofuscinosis. *Developmental Medicine and Child Neurology*, *48*, 259–264.
- Ainsworth, M. D. S. (1963). The development of infant-mother interaction among the Ganda. In B. M. Foss (Ed.), *Determinants of infant behavior, Volume 2* (pp. 67–104). New York, NY: Wiley.
- Alon, L., Ophir, M. C., Cohen, A., & Tirosh, E. (2010). Regulation disorders among children with visual impairment: A controlled study. *Journal of Developmental and Physical Disabilities*, *22*, 57–64.
- American Psychiatric Association (2013). *Diagnostic and Statistical Manual of Mental Disorders, Fifth edition (DSM-5)*. Washington, DC: American Psychiatric Association.
- Bäckman, M. L., Santavuori, P. R., Åberg, L. E., & Aronen, E. T. (2005). Psychiatric symptoms of children and adolescents with juvenile neuronal ceroid lipofuscinosis. *Journal of Intellectual Disability Research*, *49*, 25–32.
- Badrakalimuthu, V. R., & Tarbuck, A. F. (2012). Anxiety: A hidden element in dementia. *Advances in Psychiatric Treatment*, *18*, 119–128.
- Ballard, C., Day, S., Sharp, S., Wing, G., & Sorensen, S. (2008). Neuropsychiatric symptoms in dementia: Importance and treatment considerations. *International Review of Psychiatry*, *20*, 396–404.
- Bowlby, J. (1969). *Attachment and loss, Volume 1. Attachment*. Harmondsworth: Penguin.
- Bowlby, J. (1982). Attachment and loss: Retrospect and prospect. *American Journal of Orthopsychiatry*, *52*, 664–678.
- Breuer, J., & Elson, M. (2017). Frustration-aggression theory. In P. Sturmey (Ed.), *The Wiley handbook of violence and aggression* (Chapter 40, pp. 1–12). London, UK: Wiley.
- Bruininks, R. H., Woodcock, R. W., Weatherman, R. E., & Hill, B. K. (1996). *Scales of Independent Behavior – Revised: Comprehensive Manual*. Itasca, IL: Riverside.
- Cerejeira, J., Lagarto, L., & Mukaetova-Ladinska, E. (2012). Behavioral and psychological symptoms of dementia. *Frontiers in Neurology*, *3*, 73.
- Enache, D., Winblad, B., & Aarsland, D. (2011). Depression in dementia: Epidemiology, mechanisms, and treatment. *Current Opinion in Psychiatry*, *24*, 461–472.
- Goldstein, S. & Brooks, R. B. (Eds.) (2013). *Handbook of resilience in children, Second edition*. New York, NY: Springer.
- Goyal, A. R., Engedal, K., & Eriksen, S. (2019). Clinicians' experiences of anxiety in patients with dementia. *Dementia*, *18*, 80–93.
- Hankin, B. L. (2012). Future directions in vulnerability to depression among youth: Integrating risk factors and processes across multiple levels of analysis. *Journal of Clinical Child and Adolescent Psychology*, *41*, 695–718.

- Hankin, B. L., Gibb, B. E., Abela, J. R., & Flory, K. (2010). Selective attention to affective stimuli and clinical depression among youths: Role of anxiety and specificity of emotion. *Journal of Abnormal Psychology, 119*, 491–501.
- Hankin, B. L., Young, J. F., Gallop, R., & Garber, J. (2018). Cognitive and interpersonal vulnerabilities to adolescent depression: Classification of risk profiles for a personalized prevention approach. *Journal of Abnormal Child Psychology, 46*, 1521–15133.
- Harter, S. (1987). The determinants and mediation role of global self-worth in children. In N. Eisenberg (Ed.), *Contemporary issues in developmental psychology* (pp. 219–242). New York, NY: Wiley.
- Janssen, C. G. C., Schuengel, C., & Stolk, J. (2002). Understanding challenging behavior in people with severe and profound intellectual disability: A stress-attachment model. *Journal of Intellectual Disability Research, 46*, 445–453.
- Joshi, G., Petty, C., Wozniak, J., Henin, A., Fried, R., Galdo, M., Kotarski, M., Walls, S., & Biederman, J. (2010). The heavy burden of psychiatric comorbidity in youth with autism spectrum disorders: A large comparative study of a psychiatrically referred population. *Journal of Autism and Developmental Disorders, 40*, 1361–1370.
- Kerns, C. M., & Kendall, P. C. (2014). Autism and anxiety: Overlap, similarities, and differences. In T. E. Davis III, S. W. White & T. H. Ollendick (Eds), *Handbook of autism and anxiety* (pp. 75–89). New York, NY: Springer.
- Laugesen, N., Dugas, M. J., & Bukowski, W. M. (2003). Understanding adolescent worry: The application of a cognitive model. *Journal of Abnormal Child Psychology, 31*, 55–64.
- Lyketsos, C. G., & Olin, J. (2002). Depression in Alzheimer's disease: Overview and treatment. *Biological Psychiatry, 52*, 243–252.
- Marshall, F. J., de Blicke, E. A., Mink, J. W., Dure, L., Adams, H., Messing, S., et al. (2005). A clinical rating scale for Batten disease reliable and relevant for clinical trials. *Neurology, 65*, 275–279.
- Masten, A. S. (2016). Resilience in developing systems: The promise of integrated approaches. *European Journal of Developmental Psychology, 13*, 297–312
- Myrbakk, E., & von Tetzchner, S. (2008). Psychiatric disorders and behavior problems in people with intellectual disability. *Research in Developmental Disabilities, 29*, 316–332.
- Perry, E., & Flood, A. (2016). Autism spectrum disorder and attachment: A clinician's perspective. In H. K. Fletcher, A. Flood & D. J. Hare (Eds.), *Attachment in intellectual and developmental disability: A clinician's guide to practice and research* (pp. 79–103). Oxford, UK: Wiley.
- Roberts, C. (2015). Depression. In T. Gullotta, R. Plant & M. Evans (Eds.), *Handbook of adolescent behavioral problems, Second edition* (pp. 173–191). Boston, MA: Springer.
- Rubin, K. H., Bowker, J. C., McDonald, K. L., & Menzer, M. (2013). Peer relationships in childhood. In P. D. Zelazo (Ed.), *The Oxford handbook of developmental psychology, Volume 2* (pp. 242–275). New York: Oxford University Press.
- Runjic, T., Prcic, A. B., & Alimovic, S. (2015). The relationship between social skills and behavioral problems in children with visual impairment. *Hrvatska Revija za Rehabilitacijska Istrazivanja, 51*, 64–76.
- Rutter, M. (2013). Annual research review: Resilience – clinical implications. *Journal of Child Psychology and Psychiatry, 54*, 474–487.
- Sameroff, A. J. (2010). A unified theory of development: A dialectic integration of nature and nurture. *Child Development, 81*, 6–22.

- Santavuori, P., Linnankivi, T., Jaeken, J., Vanhanen, S.-L., Telakivi, T., & Heiskala, H. (1993). Psychological symptoms and sleep disturbances in neuronal ceroid-lipofuscinoses. *Journal of Inherited Metabolic Diseases*, *16*, 245–248.
- Steinberg, M., Shao, H., Zandi, P., Lyketsos, C. G., Welsh-Bohmer, K. A., Norton, M. C., Breitner, J. C. S., Steffens, D. C., & Tschanz, J. T. (2008). Point and 5-year period prevalence of neuropsychiatric symptoms in dementia: The Cache County Study. *International Journal of Geriatric Psychiatry*, *23*, 170–177.
- von Tetzchner, S. (2004). Early intervention and prevention of challenging behaviour in children with learning disabilities. *Perspectives in Education*, *22*, 85–100.
- von Tetzchner, S. (2019). *Child and adolescent psychology: Typical and atypical development*. Abingdon, UK: Routledge.
- von Tetzchner, S., Fosse, P., & Elmerskog B. (2013). Juvenile neuronal ceroid lipofuscinosis and education. *Biochimica et Biophysica Acta*, *1832*, 1894–1905.
- Zeman, J., Cassano, M., Perry-Parrish, C., & Stegall, S. (2006). Emotion regulation in children and adolescents. *Journal of Developmental and Behavioral Pediatrics*, *27*, 155–168.

Afterword

The aim of the project and this book has been to collect information about the developmental course of juvenile neuronal ceroid lipofuscinosis (JNCL) and the experiences of parents and professionals related to the education and support for children, adolescents and adults with this disease. An important motivation was to make readers aware of educational tools and strategies that may be useful but also to inspire new thoughts and insights in the readers. If that is the case, our work in this project and in editing the book has filled its purpose.

Working with individuals with JNCL is challenging, but for most of us, it is also a rewarding experience. We believe that drawing on the ideas and competence summarized in this book, readers will have a more solid basis for their work and guidance in choosing strategies which have been found to produce positive results in the past.

The project and this book would not have been possible without the formidable contributions from members of this project, parents, teachers and care givers, who have given their ideas and knowledge to the project. In addition, valuable contributions have been received from professionals with many years of experience working within the fields of education, communication and language, habilitation, blindness, dementia, music and physical therapy.

For us, the Editors, the project period (2014–2018) has been a time of learning. Many of the views expressed in this book result from long and fruitful discussions and integrate quite different points of view. No doubt, this process has added considerable value to the final product.

The main idea that JNCL must be understood from the perspectives of blindness and childhood dementia underlies the survey and interviews in the project and is very visible in the book. These two features are characteristics of the developmental course of JNCL, but many of the disease's developmental consequences are also found in people with other diseases. We believe the book will be valuable for these groups as well.

Finally, we have chosen to examine education and learning in individuals with JNCL from a lifetime perspective. This approach implies that the capacity for learning extends beyond the school years. Early intervention and the acquisition of skills, the maintenance of these skills, and ongoing learning thus constitute a continuous process, which will contribute to the quality of life also for adults with JNCL.

The Editors

Appendix A

Methods

*Bengt Elmerskog, Stephen von Tetzchner,
Anne-Grethe Tøssebro and Svein Rokne*

The aim of the present study was to investigate the experiences of parents, educational staff and care staff related to education and support services for children, adolescents and adults with juvenile neuronal ceroid lipofuscinosis (JNCL). It is a survey and interview study (see Robson & McCartan, 2016) with informants from six countries: Denmark, Finland, Germany, Norway, the UK (England and Scotland only), and the USA.

The study is a part of the international project, «Juvenile Neuronal Ceroid Lipofuscinosis (JNCL) and Education» (2014–2017) which was funded by the European Commission through Erasmus+ grant No.2014-1-NO01-KA200-000388. In addition, the project was supported by participating organizations and volunteer work. Preparatory activities in 2013 and 2014 were funded by The Directorate for Children, Youth and Family Affairs and the Directorate for Education and Training in Norway, the Norwegian NCL Family Association and Statped, Norway. Statped Midt in Norway coordinated the project.

Participants

Recruitment of informants was made through parent associations, educational institutions and medical institutions participating in the project. They distributed a letter with information about the project and a consent form to families with a member with JNCL. Parents who had given consent were asked to nominate teachers or other non-medical staff as potential informants (hereafter named "professionals"). The information letter and consent form were sent to the nominated professionals, including the information that they had been nominated by parents. Many of the parents informed the teachers or staff members that they would receive a letter of information asking them to take part in the survey. Some parents did not want to involve a teacher or another professional. Other parents did

not want to take part in the survey but did nominate a teacher or staff member who consented to take part in the project. This means that the group of individuals with JNCL described in the surveys by parents was not identical to the group described by the professionals. It is not possible to compare responses from parents and professionals, because many participants were only represented either by parents or by professionals. The reason for including information from both parents and professionals was to obtain as much information as possible and from more than one source: both a parent perspective and a professional perspective. However, it was not an aim to compare the experiences and views of parents and professionals.

The parents and professionals who completed the questionnaires and took part in interviews are here called *informants*, while the individuals with JNCL who were described are called *participants*. To protect personal information, the consent form was returned to the national coordinator of each country who de-identified the informants and participants (removed names and identifying information) before the results were sent to the researchers. The researchers thus do not know the names of those who participated.

A few teachers and other staff members completed surveys describing the same individual with JNCL, and a few teachers and staff included more than one individual in their questionnaire responses. These were not included in the data analyses presented in this book because the surveys were about individuals and not about groups.

Figure 1 shows the numbers of parents and professionals who were informants in the surveys and interviews. The number of informants in each country is not proportional to the country's population. This may reflect national differences in

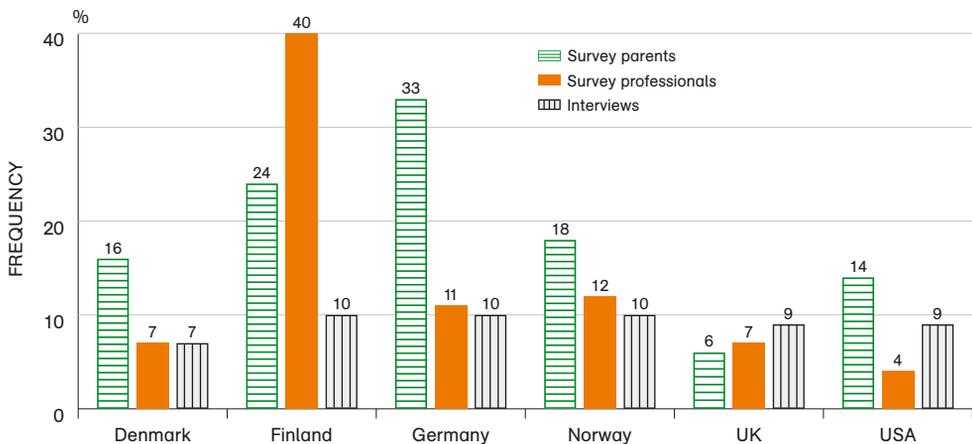


Figure A.1 The number of individuals from each country who completed questionnaires and participated in the interviews

Table A.1 Age (years) and sex distribution among participants with JNCL represented in the three surveys

Survey	N	Mean age (SD)	Range	Males/ Females
1 Parents	78	17.1 (6,5)	6–34	37/41
2 Bereaved parents	33	24.9 (5,3)	17–34	12/21
3 Professionals	81	13.0 (8.1)	8–27	41/40

Note: A few informants did not provide information about the age of the participant. In Survey 2, the mean age refers to the age when the participants passed away. The total number of participants are 135. Some of these participants appear in two different Surveys; in Survey 1 and 3 or in Survey 2 and 3.

prevalence but is mainly due to differences in the way informants were recruited. In Denmark and Norway, all families with a family member with JNCL were invited to participate in the survey through the national family organization and the educational centers responsible for the follow up of individuals with JNCL. Together they knew the names of all or nearly all families in the country. Information letters were sent to all the families, also the bereaved families. In Finland, Germany, the UK and the USA, the associations sent the information letter to families who were receiving services from the association or who had received such services in the past, including some parents who were bereaved. In Finland, most relevant families were known to the parent association. The German sample was mainly recruited from the northern part of Germany, where most families were known to the institutions participating in the project, and the sample was thus representative for this geographical part of Germany. There was a relatively low number of participants from the UK and USA. Recruitment of professionals working with individuals with JNCL depended on a nomination from parents, and thus reflects the parent group. It is not clear how these recruitment procedures have influenced the results. It is a strength that the study has a relatively large sample, but the sample is more representative of individuals with JNCL and educational systems and support services in Northern Europe than in the UK and USA.

Three different surveys were organized. Survey 1 was for parents, Survey 2 was for bereaved parents and Survey 3 was for professionals (see below). A total of 192 questionnaires were completed in the three surveys, for 135 participants, 63 males and 72 females with JNCL. Survey 1 includes information about 78 participants with an average age of 17.1 years (Table A.1). Survey 2 comprises 33 individuals who had passed away and their mean length of life was 24.9 years. Survey 3 comprises 81 participants with a mean age of 16.4 years. All participants had a diagnosis of JNCL. The youngest participant in the surveys was six and the oldest was 34 years old. All participants attended or had attended school but there

were rather few students aged between six and nine years. In surveys 1 and 3, thirty-nine participants were past school age, 20 years or older.

The plan was to complete ten interviews in each country, five with parents and five with professionals. Twenty-five interviews with parents and 30 interviews with professionals were completed, resulting in more than 2000 transcribed pages. The transcriptions of non-English interviews were translated to English. All personal names used in the interviews were de-identified in the transcriptions.

One reason for including several countries in the study was the possibility of getting a sample that was large enough to uncover the variation that may exist in development, learning and mastering among individuals with JNCL, as well as in educational systems and support services. The variation in results might reflect both biological differences and cultural difference in education and support services. The present study presents new information because it is the first large study addressing issues related to education and support for individuals with JNCL. However, this study should be supplemented with new studies that have samples that are representative for countries with other educational and support services than those in Northern Europe, which constitute the majority of the participants presented in this study.

Surveys and interviews

There were three different surveys in five different languages (Danish, English, Finnish, German and Norwegian).

- Survey 1 – For parents (with focus on the history and the present)
- Survey 2 – For bereaved parents (with focus on the history)
- Survey 3 – For teachers and other professionals (with focus on the present)

The surveys include demographic information, and information about the present status of the participants with JNCL. They include many of the same themes, but the information was somewhat different. The main parent survey includes both retrospective information and information about the present situation of the individual with JNCL, while the bereaved parents were asked to give retrospective information. The professionals were mainly asked about the present situation of the participants.

The surveys include information about when different problems became noticeable, the diagnostic process, abilities and challenges, school attendance and educational strategies, support services, and the participant's social interaction with peers. For many issues, the parents were asked to use a five-point scale to rate their

child's skills and achievements at different age levels: 1) no competence, 2) low, 3) moderate, 4) high, and 5) very high competence. Satisfaction with education was evaluated on a similar scale: 1) no satisfaction, 2) low, 3) moderate, 4) high, and 5) very high satisfaction. The participants' interests in different activities compared to peers were indicated on this scale: 1) much lower interest, 2) lower, 3) same, 4) greater, and 5) much greater interest than peers. Social interaction with peers was indicated on the scale: 1) no interaction, 2) minimal, 3) reasonable, 4) pretty good, and 5) very good interaction. For some activities and behaviors, informants were asked about frequency: 1) never, 2) monthly, 3) weekly, 4) daily, and 5) several times per day. Different moods were indicated as being present: 1) never, 2) seldom, 3) sometimes, 4) often, or 5) very often. The contributions of diverse services were evaluated on the scale: 1) no contribution, 2) some contribution, 3) moderate degree of contribution, 4) high degree of contribution, and 5) very high degree of contribution. Negative effects were indicated on a similar scale: 1) no negative effect, 2) some, 3) moderate, 4) high, and 5) very high negative effect. Informants were asked to evaluate the impact of education and services on the scale: 1) no impact, 2) low, 3) some, 4) high, and 5) very high impact. Parents indicated level of cooperation with different services on the scale: 1) no cooperation, 2) low degree of cooperation, 3) moderate, 4) high, and 5) very high degree of cooperation. Parents were asked to evaluate the usefulness of different sources of information, including from school, courses and associations on the scale: 1) not useful, 2) useful to a low degree, 3) useful to a moderate degree, 4) very useful, and 5) extremely useful.

For most issues, the informants could add comments to their evaluation scores, and many informants used this opportunity to give supplementary information and clarifying examples and views.

The interviews were semi-structured. There were two different interview guides, one for interviews with parents and one for interviews with professionals, and they were available in five languages (Danish, English, Finnish, German and Norwegian). The interviews were mostly made face-to-face but due to geographical distances and difficulties finding an appropriate time, telephone and Skype were used for a few interviews. The interviews followed the interview guide but with flexibility, and informants were free to convey any information they thought would be relevant. The interviews were conducted by local project partners and it was ensured that the interviewer was not affiliated with the informant and was not responsible for educational or other support services for the participant with JNCL. The local interviewer transcribed the interview, translated it into English when needed, and sent it to the researchers.

Informants from the surveys and interviews were de-identified and given a project identification number. Both quantitative and qualitative data were mainly analyzed in Norway.

Procedures

The questionnaire with factual information and evaluation scales was sent to all parents who had given consent. The parents were always recruited first, and professionals were only approached if parents had given consent and nominated a professional. The information letters and consent forms for teachers and other professionals were sent to the professionals indicated by the parents. Parents and professionals could complete the questionnaire on paper and send it by mail, or electronically on Internet.

The participants were asked to indicate on the questionnaire whether they were willing to participate in an in-depth interview. The parents and professionals who were interviewed were drawn among those who had expressed willingness to take part in an interview. After completion of the interview, the informants were contacted on telephone to inquire if there was anything they would like to add (they had been informed about this during the interview).

Analysis

Quantitative analyses included mainly descriptive statistics, with frequencies, mean, standard deviation and range, and a small number of correlations. All statistics were performed with IBM SPSS Version 25. Qualitative data were analyzed using a thematic approach (see Guest, MacQueen, & Namey, 2012). Direct quotations from parents' and professionals' contributions were used to illustrate trends in comments as well as trends in quantitative data.

Ethics

The study was approved by the Regional Committees for Medical and Health Research Ethics in 2015 (No. 2015/1464). The parents had control over the recruitment of professionals, as professionals were only approached if parents had given their consent and nominated a person.

References

- Guest, G., MacQueen, K. M., & Namey, E. E. (2012). *Applied thematic analysis*. London, UK: Sage.
- Robson, C., & McCartan, K. (2016). *Real world research, Fourth edition*. Chichester, UK: Wiley.

Appendix B

Resources

- Batten Disease Family Association (BDFA)
<http://www.bdfa-uk.org.uk/>
- Batten Disease Support and Research Association (BDSRA)
<https://bdsra.org>
- Beyond Batten Disease Foundation
<https://beyondbatten.org/research/state-of-science/>
- Bildungszentrum für Blinde und Sehbehinderte in Hamburg
(Educational Center for the Blind and Visually Impaired in Hamburg)
<http://bzbs.hamburg.de>
- NCL Danmark (The Danish NCL Family Association)
<http://dsvf.dk>
- NCL Gruppe Deutschland e. V. (The German NCL Family Association)
<http://www.ncl-deutschland.de>
- NCL Net
<http://ncl-netz.de/en/specialclinic.htm>
- NCL Resource
<http://www.ucl.ac.uk/ncl/>
- Norsk Spielmeyer-Vogt Forening (The Norwegian NCL Family Association)
<https://www.nsvf.org/>
- Oppimis-ja ohjauskeskus Valteri Onerva
(Learning and Consulting Center Valteri Onerva)
<https://www.valteri.fi/onerva>
- Royal Blind School
<https://www.royalblind.org/education>
- Statped
<http://www.statped.no/>
- Suomen JNCL-perheiden tukiydistys ry (The Finnish JNCL Association)
<https://www.jncl.fi>

Synscenter Refsnæs (Vision Center Refnæs)

<http://synref.dk/>

University of Rochester Batten Center (URBC)

<https://www.urmc.rochester.edu/neurology/batten-disease-center.aspx>

WESC Foundation – The Specialist Centre for Visual Impairment

<https://www.wescfoundation.ac.uk>

Juvenile neuronal ceroid lipofuscinosis (JNCL) is a rare neurological condition characterized by the onset of blindness and dementia in childhood, but with considerable individual differences. This book is concerned with the developmental course of the disease and educational and non-medical intervention for children and young people with this condition. The book is based on an international project on JNCL, dementia and education, and presents evidence-based practices in various areas. It gives the reader insight into educational strategies and tools which may support learning and maintenance of knowledge and skills in children and young people with JNCL, as well as the experiences of parents and staff. The text is illustrated with many small case stories.

The chapters are written by professionals and parents from different countries and give a broad knowledge foundation for planning education for students with JNCL and contributing to their learning and a meaningful life. The book is intended as a knowledge base and source of practice for parents, educators and support staff. The book focuses on JNCL and its many manifestations and symptoms, but may be useful also for professionals working with other young people with early blindness or dementia.



Erasmus+

ISBN 978-82-7083-563-8



9 788270 835638