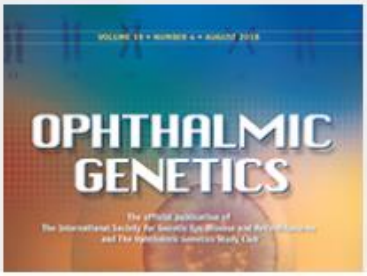

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Research Report

Cataract and Glaucoma Development in Juvenile Neuronal Ceroid Lipofuscinosis (Batten Disease)

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Abstract

Background: Ophthalmologic studies of Juvenile Neuronal Ceroid Lipofuscinosis (JNCL) have focused mainly on retinal involvement, and so far no anterior segment abnormalities have been described. In the present study, we report the findings of pre-senile cataract in five patients with JNCL.

Material and Methods: Our sample consisted of 35 patients (19 males, 16 females) with JNCL associated to the Centre for Rare Disease, Aarhus University Hospital. They represent all patients with JNCL born in Denmark in the period 1971–2003. At the half-yearly routine outpatient visits, the anterior section was examined by ordinary penlight without instillation of a mydriatic, and if abnormalities such as cataracts were detected or suspected, the patients were referred for an ophthalmologic examination including slit lamp examination. Follow up was obtained on all patients referred for ophthalmologic examination.

Results: During the study period (1996–2012), five patients were identified with cataract. The patients' average age at detection of cataract was 20.1 + 1.6 years (mean + 2SD). Two of the five patients developed acute glaucoma, and in one case prophylactic cataract surgery was performed.

Conclusions: Cataract formation and a secondary acute glaucoma are complications in JNCL which do occur. We recommend that a complete ophthalmological examination of the anterior segment should be performed routinely in patients with JNCL beyond the age of 16 years of age in order to prevent a painful and harmful acute glaucoma which may occur due to mature cataract formation.

Keywords: Cataract, neuronal ceroid lipofuscinosis, retinal degeneration, secondary glaucoma