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Therapeutic landscape for Batten disease: current treatments and future prospects.

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Abstract

Batten disease (also known as neuronal ceroid lipofuscinoses) constitutes a family of devastating lysosomal storage disorders that collectively represent the most common inherited paediatric neurodegenerative disorders worldwide. Batten disease can result from mutations in 1 of 13 genes. These mutations lead to a group of diseases with loosely overlapping symptoms and pathology. Phenotypically, patients with Batten disease have visual impairment and blindness, cognitive and motor decline, seizures and premature death. Pathologically, Batten disease is characterized by lysosomal accumulation of autofluorescent storage material, glial reactivity and neuronal loss. Substantial progress has been made towards the development of effective therapies and treatments for the multiple forms of Batten disease. In 2017, cerliponase alfa (Brineura), a tripeptidyl peptidase enzyme replacement therapy, became the first globally approved treatment for CLN2 Batten disease. Here, we provide an overview of the promising therapeutic avenues for Batten disease, highlighting current FDA-approved clinical trials and prospective future treatments.

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