

# Download Neuronal Ceroid Lipofuscinoses (batten Disease)

Neuronal ceroid lipofuscinosis (NCL) is the general name for a family of at least eight genetically separate neurodegenerative lysosomal storage diseases that result from excessive accumulation of lipopigments in the body's tissues. These lipopigments are made up of fats and proteins. Their name comes from the word stem lipo-, which is a variation on "lipid" or "fat", and from the term pigment ... Neuronal ceroid lipofuscinoses (NCLs) are a heterogeneous group of lysosomal storage disorders. NCLs include the rare autosomal recessive neurodegenerative disorder neuronal ceroid lipofuscinosis type 2 (CLN2) disease, caused by mutations in the tripeptidyl peptidase 1 (TPP1)/CLN2 gene and the resulting TPP1 enzyme deficiency. CLN2 disease most commonly presents with seizures and/or ataxia in ... What are Neuronal Ceroid Lipofuscinoses (NCLs)? Commonly referred to as Batten disease, the Neuronal Ceroid Lipofuscinoses (NCLs) denote several different genetic life-limiting neurodegenerative diseases that share similar features. The purpose of this study is to determine whether BMN 190 is safe and effective in the treatment of patients with Late-Infantile Neuronal Ceroid Lipofuscinosis Type 2 (CLN2) disease. - Neuronal Ceroid Lipofuscinoses (batten Disease)