



Need to talk? Call us 0845 2412173

Monday to Friday
9am to 5pm



Supporting those affected by
Inherited Metabolic Disorders

CLN10 Disease

CLN10 Disease is an extremely rare disorder which belongs to a group known as the Neuronal Ceroid Lipofuscinoses (NCL), previously collectively referred to as Batten Disease. They have since been renamed to reflect the causative gene and the age of onset. This form is caused by a defect in the Cathepsin D (also known as CLN10) gene. The gene is involved in the production of the cathepsin D enzyme which is present in many cells. There are two forms, depending on the amount of enzyme produced. A complete lack of enzyme causes severe neurological symptoms present at birth or earlier (Congenital CLN10 Disease). If some level of enzyme is produced symptoms become apparent later in life, often between late infancy and adulthood, with slower progression and gradual loss of skills (Late Infantile CLN10 Disease).

Synonyms

Alternative names for this condition are:

- Cathepsin D Deficiency
- Cathepsin D Deficient Neuronal Ceroid Lipofuscinosis
- CLN10
- Neuronal Ceroid Lipofuscinosis 10

Further information about this condition is available from Climb.



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