Climb™

Supporting those affected by Inherited Metabolic Disorders

CLN1 Disease, Infantile

CLN1 Disease belongs to a group known as the Neuronal Ceroid Lipofuscinoses, 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 CLN1 gene which provides instructions for the PPT1 protein. A deficiency of this protein causes problems in the storage of proteins and certain fats in the nerve cells and other cells leaving them unable to work properly and leading to the symptoms of this disorder. As determined by the name, symptoms begin in infancy although rarely CLN1 may present as a late-infantile, juvenile, or adulthood onset. Symptoms are progressive and primarily affect the nervous system.

Synonyms

Alternative names for this condition are:

- Infantile Batten Disease
- Infantile CLN1 Disease
- Santavuori-Haltia Disease

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

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