Climb™

Supporting those affected by Inherited Metabolic Disorders

CLN6 Disease, Variant Late-Infantile

CLN6 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 CLN6 gene which provides instructions for a protein found in a structure within the cell which is involved in processing and transporting proteins. It is believed to help the cell to get rid of substances that are no longer needed. The gene defect causes an abnormal form of the protein to be produced which is broken down quickly, meaning there isn't enough working protein in the cell. This leads to the symptoms of this disorder. Symptoms usually begin between early and late childhood with delayed development followed by the onset of seizures. In some cases, symptoms begin in adulthood, usually after age 30 presenting with epilepsy and a progressive decline of neurological function. Symptoms are progressive and primarily affect the nervous system.

Synonyms

Alternative names for this condition are:

- Early Juvenile Batten Disease

Further information about this condition is available from Climb.

Disclaimer

Updated 03/08/17

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