Climb™

Supporting those affected by Inherited Metabolic Disorders

CLN8 Disease, Variant Late-Infantile

CLN8 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 CLN8 gene which provides instructions for a protein believed to assist a structure within the cell which is involved in processing and transporting proteins around the cells. CLN8 Disease causes a deficiency or production of an impaired version of the CLN8 protein causing the cells not to function properly. This leads to the symptoms of this disorder. There are 2 forms of this disorder - the lesser severe form is characterised by epilepsy from approximately 5-10 years of age and is commonly known as Northern Epilepsy as it appears to affect only those with ancestry from Northern Finland. The more severe form is characterised by the onset of seizures between ages 2 and 7 followed by progressive neurological symptoms.

Synonyms

Alternative names for this condition are:

- Variant Late-Infantile Batten Disease
- Variant Late Infantile Neuronal Ceroid Lipofuscinosis
- Epilepsy with Progressive Mental Retardation (EPMR)
- Northern Epilepsy

Further information about this condition is available from Climb.

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