Climb™

Supporting those affected by Inherited Metabolic Disorders

CLN7 Disease, Variant Late-Infantile

CLN7 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 CLN7 gene which provides instructions for the MFSD8 protein which is believed to transport molecules across small membranes found inside the cell. It is not yet understood how this leads to the symptoms of this disorder. Symptoms usually begin between the ages of 2 and 7 with epilepsy and a loss of skills. Symptoms are progressive and primarily affect the nervous system.

Synonyms

Alternative names for this condition are:

- Variant Late-Infantile Batten Disease

Further information about this condition is available from Climb.

Disclaimer

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