Climb™

Supporting those affected by Inherited Metabolic Disorders

CLN5 Disease, Variant Late-Infantile

CLN5 Disease 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 CLN5 gene. Although it is not known which specific protein the CLN5 gene provides instructions for, a number of different mutations of the CLN5 gene have been linked to this form of NCL; all of which cause 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. Symptoms usually become noticeable at school age with slowed development and the onset of behavioural problems. Epilepsy usually becomes evident between ages 7 and 13. Symptoms are progressive and primarily affect the nervous system.

Synonyms

Alternative names for this condition are:

- Variant Late-Infantile Batten Disease
- Variant Late-Infantile CLN5 Disease
- Variant Late-Infantile Neuronal Ceroid Lipofuscinosis

Further information about this condition is available from Climb.

Disclaimer

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Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: www.climb.org.uk