Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

Niemann-Pick Disease Type C

This condition belongs to the Niemann-Pick group of disorders where there are difficulties in breaking down fats. Type C is a neurodegenerative disorder characterised by an enlargement of the liver and spleen, yellowing of the skin and eyes, and a loss of a certain type of eye movement. Niemann-Pick Disease Type C is caused by a unique error in the cellular trafficking of cholesterol and other fatty acids, so the lysosomes, a part of the cell, accumulate cholesterol.

The onset of symptoms can present at any time, in the foetal period through to adulthood. All children and adults are different and the symptoms are extremely variable. Symptoms may primarily involve the liver or involve neurological or psychiatric problems. In 95% of cases, this disorder is caused by a defect in the NPC1 gene. In the remaining 5% of families it is caused by a different gene, NPC2.

Synonyms

Alternative names for this condition are:

- NP-C

Further information about this condition is available from Climb.

Disclaimer

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