

Niemann-Pick disease type C (NPC) is a very rare, inherited neurodegenerative disease that results from an abnormal processing in body tissues of fatty substances (lipids), particularly cholesterol. Fatty substances accumulate in the brain, spleen, liver and lung, causing dementia and other symptoms leading to disability and premature death. NPC is one of many rare hereditary disorders of the metabolism of lipids and other molecules that result in a neurodegenerative disease.

About Niemann-Pick disease type C

NPC is an inherited neurodegenerative disease that causes increased damage to the nervous system over time. This condition is referred to as a “lipid storage disease” that is caused by the body’s inability to process and remove abnormal amounts of cholesterol or other lipids (fats) from the body.

With the body being unable to properly breakdown these fats, an abnormally large cholesterol buildup will develop in the spleen and liver, while harmful amounts of other lipids begin to collect in the brain. As a result of the damaging accumulation of lipids throughout the body, NPC may affect the physical, neurological, emotional and cognitive functioning of people with this rare disease.

What are the symptoms?

Symptoms may begin at any age, most commonly in school age children; however, infants, adolescents and adults can all be affected by the disease. With greater awareness and better diagnostic methods, more patients are being identified in adulthood. Progression is more rapid with earlier onset.

Childhood symptoms of NPC may be first noticed when a child starts going to school and concerns regarding a child’s behaviour are raised. As the disease progresses, children may experience developmental delays, challenges with motor skills (e.g. increased clumsiness and abnormal posturing) and a particular difficulty in making vertical eye movements. Other symptoms of NPC may include: an enlarged liver or spleen, challenges with swallowing, slurred speech, loss of muscle strength, seizures, breathing problems and gradual liver failure. Another particular symptom is episodes of falling with laughter called “gelastic cataplexy” due to a sudden loss of muscle tone.

People who begin to show signs of NPC in their early adulthood are more likely to develop symptoms of dementia than those people who develop NPC at a younger age. The dementia-related symptoms are characterized by initial impairments in executive functions such as dis-inhibition, inflexibility of thinking, poor judgement, lack of insight, inability to understand abstract concepts; poor attention and mental slowing followed by increasing difficulty with short-term memory and learning.

How is Niemann-Pick disease type C diagnosed?

If a person has the clinical features of NPC, neurologists or specialists in genetic diseases will likely perform a skin biopsy. This procedure requires the removal of a small piece of skin that is sent to a special laboratory where cells are cultured and then tested for the capacity to process cholesterol. A new diagnostic test that requires blood only measures the products of cholesterol processing.

DNA testing may also be recommended if the cultured skin cells reveal abnormal processing of cholesterol. A DNA test will look for mutations in one of two genes (NPC1 and NPC2) that could be causing abnormal function of proteins involved in processing of cholesterol and other lipids.

What are the causes or the risk factors?

NPC is an inherited disease caused by the body's inability to properly metabolize (break down) cholesterol and lipids. This can lead to harmful amounts of cholesterol buildup in the spleen and liver, as well as an accumulation of fat in the brain that can cause a progressive decline in functioning. This lack of ability to process cholesterol in the body is the result of a genetic mutation in either the NPC1 or NPC2 gene.

Is there treatment?

There is currently no cure for NPC; however, there is now a drug called Miglustat that can prevent the body from producing excess lipids and delays progression of the disease. Thus early diagnosis has become important. Other medications can help manage some of the symptoms caused by NPC, such as seizures, but they cannot stop the progression of the disease. Research on potential treatments has been moving forward since 1997 when scientists identified the NPC1 gene involved in NPC.

Therapeutic approaches are also supporting people living with NPC to manage symptoms of their disease. Occupational and physical therapies may help improve the functional ability of people living with NPC through exercises that are focused on movement and improving muscle strength. Speech therapy can help improve verbal communication and can also address any swallowing issues that may result from the progression of NPC.

Support is available:

Visit the Alzheimer Society's website at www.alzheimer.ca or contact your local Alzheimer Society.

Additional resources:

- Canadian Chapter of the National Niemann-Pick Disease Foundation Inc.: www.nnpdf.ca
- Alzheimer's Society UK: www.alzheimers.org.uk/site/scripts/documents_info.php?documentID=135

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Sources:

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