Niemann-Pick Disease Type A/B

What Your Results Mean

Test results indicate that you are a carrier of Niemann-Pick disease type A/B. Carriers typically show no symptoms. You and your partner would both have to be carriers of Niemann-Pick disease type A/B for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner/donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner/donor are carriers for Niemann-Pick disease type A/B, each of your children has a 1 in 4 (25%) chance to have the condition.

Niemann-Pick Disease Type A/B Explained

What is Niemann-Pick Disease Type A/B?

Niemann-Pick disease type A/B is an inherited metabolic disorder where there is a defect in the enzyme called acid sphingomyelinase, which processes lipids (fats). As a result, affected individuals have accumulations of sphingomyelin, cholesterol, and other lipids in the cells of the liver, spleen, bone marrow, lungs, and central nervous system. Pathogenic variants in the *SMPD1* gene can cause either type A or type B Niemann-Pick disease.

Niemann-Pick disease type A causes intellectual disability, loss of motor skills, and enlargement of the liver and spleen, among other symptoms. Symptoms usually begin within the first few months of life, and the disease is often fatal by the age of two or three. Difficulty feeding, enlarged abdomen, developmental regression, seizures, and spastic movements are common. Most individuals will not learn to sit independently, crawl, or walk.

Unlike type A, which is fatal in early childhood, people with Niemann-Pick disease type B have a less severe course of the disease and often live into adulthood. Symptoms can begin in late childhood or adolescence. The most common symptoms include an enlargement of the liver and spleen (hepatosplenomegaly), a progressive decline in lung function, repeated respiratory infection, and poor or slower physical growth leading to shorter stature, coronary artery disease, and a decreased number of blood platelets.

Prognosis

Prognosis is generally unfavorable. The type A form of this disease is progressively neurodegenerative and usually leads to death within the first three years of life. For the type B form, life expectancy is reduced. Individuals with the type B form who survive early childhood can have progressive and/or clinically significant neurologic manifestations. Those who further survive into adulthood are at increased risk of heart disease and worsening lung function.

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 7/2020

Treatment

There is no cure for Niemann-Pick disease type A, and treatment is supportive. Bone marrow transplantation has been used with limited success and does not address the neurological symptoms. Physical and occupational therapy can maximize range of motion and avoid contractures that restrict joint movement.

There is no cure for Niemann-Pick disease type B. Treatment is supportive, involving splenectomy, supplemental oxygen, and close monitoring of calorie intake.

Resources

International Center for Types A and B Niemann-Pick Disease
http://icahn.mssm.edu/research/niemann
National Niemann-Pick Disease Foundation
https://nnpdf.org/
National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

Revised: 7/2020