Niemann-Pick Disease Type C, *NPC1*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Niemann-Pick disease type C, *NPC1*-related. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Niemann-Pick disease type C, *NPC1*-related 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 or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for Niemann-Pick disease type C, *NPC1*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Niemann-Pick Disease Type C, *NPC1*-Related Explained

What is Niemann-Pick Disease Type C, *NPC1*-Related?

Niemann-Pick disease type C, *NPC1*-related is an inherited lysosomal storage disorder that impairs fat metabolism causing harmful amounts of lipids to accumulate in the spleen, liver, lungs, bone marrow, and brain. Symptoms typically appear in childhood, although infant and adult onsets are possible. Signs of Niemann-Pick disease include severe liver disease, breathing difficulties, developmental delays, seizures, poor muscle tone, lack of coordination, problems with feeding, and an inability to move the eyes vertically.

Prognosis

Prognosis is generally poor, as affected individuals are only expected to survive for 10-20 years after diagnosis.

Treatment

There is no cure for Niemann-Pick disease type C, *NPC1*-related; however, seizure medications and physical and speech therapy may be recommended.

Resources

National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/niemann-pick-disease-type-c/

Niemann-Pick Disease Foundation
https://nnpdf.org/overview/

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