Genetics Uncoded:

NxGen MDx

Facts about

Niemann-Pick Disease (SMPD1-Associated)

What Your Test Results Mean

Carriers of *SMPD1*-associated Niemann-Pick Disease (NPD), also referred to as acid sphingomyelinase deficiency, typically show no symptoms; however, carriers are at an increased risk of having a child with NPD. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Niemann-Pick Disease Explained

NPD is an inherited metabolic disorder caused by the inability to break down fats in the brain and nervous system. In the more severe form, type A, an enlarged liver and spleen, lung disease, weakness, and developmental regression typically occur within the first six months of life with death between ages three to four years. Type B is a milder, later onset form that does not often affect the brain. Survival into adulthood is possible with NPD type B.

Individuals with NPD do not produce enough of one of the enzymes, acid sphingomyelinase or ASM, needed to metabolize the fatty substance sphingomyelin. Over time, excessive sphingomyelin builds up in the lysosomes, or recycling departments, of the cells. This excessive storage of sphingomyelin can cause permanent cellular and tissue damage, particularly in the spleen, liver, bone marrow, lungs, and sometimes the brain. Treatment of individuals with type A is symptomatic. Variable results have been reported with stem cell transplant with some reduction in liver and spleen volumes; however, neurological symptoms seen in type A are not corrected. Clinical trials are currently underway for enzyme replacement therapy for individuals with NPD type B.

How the Genetics Work

The clinical features of NPD can be explained by variants in the *SMPD1* gene. In general, individuals have two copies of the *SMPD1* gene. Carriers of NPD have a single variant in one copy of the *SMPD1* gene while individuals with NPD have variants in both copies of their genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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