Genetics Uncoded:

NxGen MDx

Facts about Gaucher Disease

What Your Test Results Mean

Carriers show no symptoms of Gaucher disease; however, some carriers have been reported to be at an increased risk for developing Parkinson's disease. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Gaucher Disease Explained

Gaucher disease is an inherited metabolic disorder in which harmful amounts of a sphingolipid called glucocerebroside accumulate within lysosomes, or recycling compartments, of cells. Individuals with Gaucher disease do not produce enough of one of the enzymes (β-glucosidase or glucocerebrosidase) needed to metabolize glucocerebroside. Over time, this excessive storage in the lysosomes can cause permanent cellular and tissue damage, particularly in the spleen, liver, bone marrow, and although rarely, the brain.

There are three forms of Gaucher disease. In the most common form, type I, the brain is not involved. Enzyme replacement therapy is available for Gaucher disease type I to slow the accumulation of sphingolipids in the body. Due to the involvement of the brain, individuals with Gaucher disease types II and III may not benefit in the long-term from enzyme replacement therapy. Stem cell transplantation has been accomplished with variable results in patients with type III Gaucher disease.

How the Genetics Work

The clinical features of Gaucher disease can be explained by pathogenic variants in the *GBA* gene. In general, individuals have two copies of the *GBA* gene. Carriers of Gaucher disease have a single variant in one copy of the *GBA* gene while individuals with Gaucher disease 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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