Facts about Gaucher Disease

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.