Gaucher Disease

What Your Results Mean

Test results indicate that you are a carrier of Gaucher disease. Carriers are not expected to show symptoms; however, carriers have a slightly increased chance to develop Parkinson's disease later in life. You and your partner or donor would both have to be carriers of Gaucher disease 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.

We recommend that you share this information with all of your health care providers. 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 or donor are carriers for Gaucher disease, each of your children have a 1 in 4 (25%) chance to have the condition.

Gaucher Disease Explained

What is Gaucher Disease?

Gaucher disease is an inherited metabolic disorder in which harmful amounts of glucocerebroside accumulate within lysosomes, the recycling compartments in cells. Individuals with Gaucher disease do not produce enough of an enzyme known as -glucosidase or glucocerebrosidase that is needed to metabolize glucocerebroside. Over time, this excessive accumulation in the lysosomes can cause permanent cellular and tissue damage, particularly in the spleen, liver, bone marrow, and rarely, the brain.

Gaucher disease is characterized by an enlarged liver and spleen, anemia, a decrease in blood platelets, lung disease, bone abnormalities, and Parkinson's disease. There are three major types of Gaucher disease. In the most common form, type 1, the brain and spinal cord are not involved. Types 2 and 3 are called the neuronopathic forms because they also impact the central nervous system. Symptoms of types 2 and 3 also include abnormal eye movements, seizures, and brain damage. Additionally, there are two less common types of Gaucher disease known as the perinatal lethal form and the cardiovascular form.

Prognosis

Prognosis is generally unfavorable and depends on the type of Gaucher disease. Prognosis is most favorable in type 1, which constitutes >90% of cases, though life expectancy is still reduced due to heart disease, stroke, lung disease, and/or liver abnormalities. Individuals with type 2 typically die between ages 2-4 years. Type 3 is less severe, and individuals can live into their third or fourth decade. Individuals with the perinatal lethal form die in utero or shortly after birth. The prognosis for the cardiovascular form depends on the success of valve replacement surgery.

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Treatment

Enzyme replacement therapy is available for Gaucher disease type 1 to slow the accumulation of sphingolipids in the body. Due to the involvement of the brain, individuals with Gaucher disease types 2 and 3 may not benefit from enzyme replacement therapy in the long-term. Stem cell transplantation has been used with variable results to treat type 3.

Resources

National Gaucher Foundation
https://www.gaucherdisease.org
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/gaucher-disease
National Society of Genetic Counselors
https://www.nsgc.org/

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