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My46 Trait Profile

Gaucher disease

Other Names: Glucocerebrosidase deficiency, glucosylceramidase deficiency

Gaucher disease is a genetic condition characterized by the body's inability to break down a certain fatty substance called glucocerebroside. It is caused by mutations in the GBA gene that makes the beta-glucocerebrosidase protein.

Characteristics of Gaucher disease

Individuals with Gaucher disease cannot properly breakdown a certain fatty substance called glucocerebroside (GL-1). GL-1 cannot be broken down because the enzyme, beta-glucocerebrosidase, is not working properly. Without enough enzyme, GL-1 builds up in various tissues of the body. Gaucher disease is divided into 3 main types: type 1, type 2, and type 3. Type 1 is the most common and is also referred to as "non-neuronopathic" Gaucher disease. Individuals with Gaucher disease type 1 often have an enlarged spleen and liver, and bone disease. Enlargement of the spleen and liver can cause abdominal pain and in rare cases, spleen rupture or liver failure. Individuals with Gaucher disease type 1 often have pain in their joints and bones and are at increased risk for low bone mineral density, which then causes their bones to break easily. Many individuals with Gaucher disease type 1 have anemia (i.e., low blood count), and bruise very easily because their blood cells do not clot properly. They also often have lung problems (e.g., pulmonary hypertension).

In Gaucher disease type 1, the brain and spinal cord (the central nervous system) are usually not affected, whereas in the rarer types (type 2 and type 3, also called the "neuronopathic" forms of Gaucher disease), the central nervous system is affected. Type 2 and type 3 Gaucher disease typically diagnosed in early childhood (i.e., before age 2).

Diagnosis/Testing

A diagnosis of Gaucher disease can be made by measuring the beta-glucocerebrosidase enzyme activity from a blood sample. A person with Gaucher disease often has deficient (low) enzyme activity. A diagnosis of Gaucher disease can also be done by genetic testing for changes or mutations in the GBA gene. This gene makes the beta-glucocerebrosidase enzyme that is responsible for breaking down the fatty substance GL-1. Mutations in the GBA gene prevent the enzyme from working properly, resulting in a build-up of GL-1 in various organs of the body.

Management/Surveillance

Management of Gaucher disease includes routine blood tests to monitor for anemia and clotting ability, bone scans and x-rays to monitor bone disease, MRIs to monitor the spleen and liver, and regular comprehensive clinical examinations by a healthcare provider familiar with Gaucher disease.

Enzyme replacement therapy (ERT) is a medical treatment available for Gaucher disease. ERT provides the beta-glucocerebrosidase enzyme that is missing in individuals with Gaucher disease through regular intravenous (IV) infusions. ERT is a lifelong treatment for Gaucher disease and is not a cure. There is also substrate reduction therapy for Gaucher disease which involves taking a medication that is designed to reduce the amount of the fatty substance GL-1 that accumulates in a cell.

Mode of inheritance

Gaucher disease is inherited in an autosomal recessive pattern. This means that an individual has to inherit two GBA mutations (i.e., one from each parent) to be affected with Gaucher disease. If both parents are carriers of a GBA mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with Gaucher disease.

Risk to family members

Parents of a child with Gaucher disease are carriers of Gaucher disease. If a sibling of a child with Gaucher disease is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of Gaucher disease.

Special considerations

None

Resources

Genetics Home Reference: Gaucher Disease
http://ghr.nlm.nih.gov/condition/gaucher-disease
National Gaucher Foundation
http://www.gaucherdisease.org

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