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Gaucher disease

Gaucher disease is a rare genetic disorder in which a person lacks an enzyme called glucocerebrosidase (GBA).

Causes

Gaucher disease is rare in the general population. People of Eastern and Central European (Ashkenazi) Jewish heritage are more likely to have this disease.

It is an autosomal recessive disease. This means that the mother and father must both pass one variant copy of the disease gene to their child in order for the child to develop the disease. A parent who carries one variant copy of the gene, but doesn't have the disease, is called a silent carrier.

The lack of GBA causes harmful substances to build up in the liver, spleen, bones, and bone marrow. These substances prevent cells and organs from working properly.

There are three main subtypes of Gaucher disease:

- Type 1 is most common. It involves bone disease, anemia, an enlarged spleen and low platelets (thrombocytopenia). Type 1 affects both children and adults. It is most common in the Ashkenazi Jewish population.
- Type 2 usually begins in infancy with severe neurologic involvement. This form can lead to rapid, early death.
- Type 3 may cause liver, spleen, and brain problems. People with this type may live into adulthood.

Symptoms

Bleeding because of low platelet count is the most common symptom seen in Gaucher disease. Other symptoms may include:

- Bone pain and fractures
- Cognitive impairment (decreased thinking ability)
- Easy bruising
- Enlarged spleen
- Enlarged liver
- Fatigue
- Heart valve problems

- Lung disease (rare)
- Seizures
- Severe swelling at birth
- Skin changes

Exams and Tests

Your health care provider will perform a physical exam and ask about the symptoms.

The following tests may be done:

- Blood test to look for enzyme activity
- Bone marrow aspiration
- MRI
- CT
- X-ray of the skeleton
- Genetic testing

Treatment

Gaucher disease can't be cured. But treatments can help regulate and may improve symptoms.

Medicines may be given to:

- Replace the missing GBA (enzyme replacement therapy) to help reduce spleen size, bone pain, and improve thrombocytopenia.
- Limit production of fatty chemicals that build up in the body.

Other treatments include:

- Medicines for pain
- Surgery for bone and joint problems, or to remove the spleen
- Blood transfusions

Support Groups

These groups can provide more information on Gaucher disease:

- National Gaucher Foundation -- www.gaucherdisease.org [https://www.gaucherdisease.org]
- MedlinePlus - Gaucher disease.-- medlineplus.gov/genetics/condition/gaucher-disease/ [https://medlineplus.gov/genetics/condition/gaucher-disease/]
- National Organization for Rare Diseases -- rarediseases.org/rare-diseases/gaucher-disease/ [https://rarediseases.org/rare-diseases/gaucher-disease/]

Outlook (Prognosis)

How well a person does depends on their subtype of the disease. The infantile form of Gaucher disease (Type 2) may lead to early death. Most affected children die before age 5.

Adults with the type 1 form of Gaucher disease can expect a normal life expectancy with enzyme replacement therapy.

Possible Complications

Complications of Gaucher disease may include:

- Seizures
- Anemia
- Thrombocytopenia
- Bone problems

Prevention

Genetic counseling is recommended for prospective parents with a family history of Gaucher disease. Testing can determine if both parents carry one variant copy of the gene and thus could pass on the Gaucher disease. A prenatal test can also tell if a baby in the womb has Gaucher disease.

Alternative Names

Glucocerebrosidase deficiency; Glucosylceramidase deficiency; Lysosomal storage disease - Gaucher; Gaucher's disease

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Review Date 11/6/2024

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