

What is Gaucher disease?

Gaucher disease is an inherited disease characterized by bone disease, enlargement of the spleen and liver, anemia, and bleeding problems, along with nervous system involvement in some individuals. In individuals with Gaucher disease, there is a deficiency in the enzyme beta-glucocerebrosidase, which breaks down a fatty component of cells called glucocerebroside. Many of the symptoms associated with Gaucher disease are attributed to the accumulation of glucocerebroside in the liver, spleen, bone marrow, and other organs.¹

What are the symptoms of Gaucher disease and what treatment is available?

Gaucher disease is most often classified into three major types that are distinguished by the presence (or absence) and severity of primary neurological symptoms. The disease varies in severity and age of onset, even within types.^{1,2}

Type	Typical age of onset	Primary neurological symptoms	Severity
Type 1	Childhood to adulthood	None	Variable
Type 2	Infancy	Severe	Often fatal by early childhood
Type 3	Childhood to early adulthood	Chronic and progressive	Reduced life expectancy; childhood to the 30s

Other subtypes include a perinatal-lethal form associated with prenatal complications, skin manifestations, and death before or during infancy, and a cardiovascular form associated with heart and eye problems.¹

Symptoms of Gaucher disease may include:^{2, 3, 4}

- Hepatosplenomegaly (enlargement of the liver and spleen)
- Bone disease, including bone pain, decreased bone density and tendency to fracture
- Anemia and easy bruising or bleeding
- Fatigue
- Lung disease, which may include pulmonary hypertension (high blood pressure in the lungs)
- Primary neurological problems, such as seizures, swallowing problems, eye problems, and difficulties with movement (types 2 or 3)
- Increased risk of Parkinson disease or parkinsonism (type 1)
- Dry, scaly skin or other skin problems (type 2)
- Limited mental and motor development (type 2)
- Failure to thrive (poor growth and weight gain) (type 2)

Treatment for nonneurological symptoms of Gaucher disease may include enzyme replacement therapy or other medications to reduce the amount of glucocerebroside that accumulates in the body. Bone marrow transplantation may benefit individuals with chronic neurological symptoms. Removal of part (or all) of the spleen may help spleen

enlargement or bleeding problems. Supportive care may include blood transfusions for anemia and bleeding, pain control for bone disease, and calcium supplementation to maintain bone density.²

How is Gaucher disease inherited?

Gaucher disease is an autosomal recessive disease caused by mutations in the *GBA* gene.¹ An individual who inherits one gene mutation in the *GBA* gene is a carrier and is typically not expected to have related health problems. An individual who inherits two mutations in the *GBA* gene, one from each parent, is expected to be affected with Gaucher disease.

If both members of a couple are carriers of a mutation in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

Who is at risk for Gaucher disease?

Gaucher disease can occur in individuals of all races and ethnicities. In the Ashkenazi (Eastern European) Jewish population, type 1 occurs more frequently with an estimated carrier frequency of 1 in 15⁵ and a calculated disease incidence of approximately 1 in 900.

Having a relative who is a carrier or who is affected can also increase an individual's chance of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members

What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

Where can I get more information?

National Gaucher Foundation: www.gaucherdisease.org

Childrens Gauchers Disease Research Fund: www.childrengaucher.org

Gaucher Care: www.gauchercare.com/en/

References

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