What is Gaucher disease?

Gaucher disease is an inherited disorder that affects many of the body's organs and tissues. The signs and symptoms of this condition vary widely among affected individuals. Researchers have described several types of Gaucher disease based on their characteristic features.

Type 1 Gaucher disease is the most common form of this condition. Type 1 is also called non-neuronopathic Gaucher disease because the brain and spinal cord (the central nervous system) are usually not affected. The features of this condition range from mild to severe and may appear anytime from childhood to adulthood. Major signs and symptoms include enlargement of the liver and spleen (hepatosplenomegaly), a low number of red blood cells (anemia), easy bruising caused by a decrease in blood platelets (thrombocytopenia), lung disease, and bone abnormalities such as bone pain, fractures, and arthritis.

Types 2 and 3 Gaucher disease are known as neuronopathic forms of the disorder because they are characterized by problems that affect the central nervous system. In addition to the signs and symptoms described above, these conditions can cause abnormal eye movements, seizures, and brain damage. Type 2 Gaucher disease usually causes life-threatening medical problems beginning in infancy. Type 3 Gaucher disease also affects the nervous system, but it tends to worsen more slowly than type 2.

The most severe type of Gaucher disease is called the perinatal lethal form. This condition causes severe or life-threatening complications starting before birth or in infancy. Features of the perinatal lethal form can include extensive swelling caused by fluid accumulation before birth (hydrops fetalis); dry, scaly skin (ichthyosis) or other skin abnormalities; hepatosplenomegaly; distinctive facial features; and serious neurological problems. As its name indicates, most infants with the perinatal lethal form of Gaucher disease survive for only a few days after birth.

Another form of Gaucher disease is known as the cardiovascular type because it primarily affects the heart, causing the heart valves to harden (calcify). People with the cardiovascular form of Gaucher disease may also have eye abnormalities, bone disease, and mild enlargement of the spleen (splenomegaly).

How common is Gaucher disease?

What is Gaucher disease?
Gaucher disease occurs in 1 in 50,000 to 100,000 people in the general population. Type 1 is the most common form of the disorder; it occurs more frequently in people of Ashkenazi (eastern and central European) Jewish heritage than in those with other backgrounds. This form of the condition affects 1 in 500 to 1,000 people of Ashkenazi Jewish heritage. The other forms of Gaucher disease are uncommon and do not occur more frequently in people of Ashkenazi Jewish descent.

What genes are related to Gaucher disease?

Mutations in the *GBA* gene cause Gaucher disease. The *GBA* gene provides instructions for making an enzyme called beta-glucocerebrosidase. This enzyme breaks down a fatty substance called glucocerebroside into a sugar (glucose) and a simpler fat molecule (ceramide). Mutations in the *GBA* gene greatly reduce or eliminate the activity of beta-glucocerebrosidase. Without enough of this enzyme, glucocerebroside and related substances can build up to toxic levels within cells. Tissues and organs are damaged by the abnormal accumulation and storage of these substances, causing the characteristic features of Gaucher disease.

Read more about the *GBA* gene.

How do people inherit Gaucher disease?

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Where can I find information about diagnosis or management of Gaucher disease?

These resources address the diagnosis or management of Gaucher disease and may include treatment providers.

- [Baby's First Test](#)
- [Gene Review: Gaucher Disease](#)
- [Genetic Testing Registry: Gaucher disease](#)
- [MedlinePlus Encyclopedia: Gaucher Disease](#)

You might also find information on the diagnosis or management of Gaucher disease in [Educational resources](#) and [Patient support](#).

General information about the diagnosis and management of genetic conditions is available in the Handbook. Read more about [genetic testing](#), particularly the difference between [clinical tests and research tests](#).

To locate a healthcare provider, see [How can I find a genetics professional in my area?](#) in the Handbook.
Where can I find additional information about Gaucher disease?

You may find the following resources about Gaucher disease helpful. These materials are written for the general public.

- MedlinePlus - Health information (2 links)
- Genetic and Rare Diseases Information Center - Information about genetic conditions and rare diseases
- Additional NIH Resources - National Institutes of Health (3 links)
- Educational resources - Information pages (10 links)
- Patient support - For patients and families (6 links)

You may also be interested in these resources, which are designed for healthcare professionals and researchers.

- Gene Reviews - Clinical summary
- Genetic Testing Registry - Repository of genetic test information (7 links)
- ClinicalTrials.gov - Linking patients to medical research
- PubMed - Recent literature
- OMIM - Genetic disorder catalog (4 links)

What other names do people use for Gaucher disease?

- cerebroside lipidosis syndrome
- Gaucher's disease
- Gauchers disease
- Gaucher splenomegaly
- Gaucher syndrome
- GD
- glucocerebrosidase deficiency
- glucocerebrosidosis
- glucosylceramidase deficiency
- glucosylceramide beta-glucosidase deficiency
- glucosylceramide lipidosis
- glucosyl cerebroside lipidosis
- kerasin histiocytosis
- kerasin lipoidosis
- kerasin thesaurismosis
- lipoid histiocytosis (kerasin type)

For more information about naming genetic conditions, see the Genetics Home Reference Condition Naming Guidelines and How are genetic conditions and genes named? in the Handbook.

What if I still have specific questions about Gaucher disease?

Ask the Genetic and Rare Diseases Information Center.