



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).

Frequency

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.

Causes

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.

Inheritance Pattern

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.

Other Names for This Condition

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

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
[/primer/testing/genetic-testing](#)
- Genetic Testing Registry: Acute neuronopathic Gaucher's disease
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268250/>
- Genetic Testing Registry: Gaucher disease
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017205/>
- Genetic Testing Registry: Gaucher disease type 3A
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1856491/>
- Genetic Testing Registry: Gaucher disease type 3B
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1856492/>
- Genetic Testing Registry: Gaucher disease type 3C
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1856476/>
- Genetic Testing Registry: Gaucher's disease, type 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1961835/>
- Genetic Testing Registry: Subacute neuronopathic Gaucher's disease
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0268251/>

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22gaucher+disease%22>

Other Diagnosis and Management Resources

- Baby's First Test
<https://www.babysfirsttest.org/newborn-screening/conditions/gaucher>
- GeneReview: Gaucher Disease
<https://www.ncbi.nlm.nih.gov/books/NBK1269>
- MedlinePlus Encyclopedia: Gaucher Disease
<https://medlineplus.gov/ency/article/000564.htm>
- National Organization for Rare Disorders (NORD) Physician Guide
<https://rarediseases.org/physician-guide/gaucher-disease/>

Additional Information & Resources

Health Information from MedlinePlus

- Encyclopedia: Gaucher Disease
<https://medlineplus.gov/ency/article/000564.htm>
- Health Topic: Gaucher Disease
<https://medlineplus.gov/gaucherdisease.html>

Genetic and Rare Diseases Information Center

- Gaucher disease
<https://rarediseases.info.nih.gov/diseases/8233/gaucher-disease>
- Gaucher disease type 1
<https://rarediseases.info.nih.gov/diseases/2441/gaucher-disease-type-1>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Gaucher's Disease Information Sheet
<https://www.ninds.nih.gov/Disorders/All-Disorders/Gaucher-Disease-Information-Page>
- National Institute of Neurological Disorders and Stroke: Lipid Storage Diseases Fact Sheet
<https://www.ninds.nih.gov/Disorders/All-Disorders/Lipid-storage-diseases-Information-Page>
- Researching Disease: Dr. Roscoe Brady and Gaucher Disease
<https://history.nih.gov/exhibits/gaucher/>

Educational Resources

- JAMA Patient Page: Gaucher disease
<https://jamanetwork.com/journals/jama/fullarticle/208870>
- MalaCards: gaucher disease, type i
https://www.malacards.org/card/gaucher_disease_type_i
- MalaCards: gaucher disease, type ii
https://www.malacards.org/card/gaucher_disease_type_ii
- MalaCards: gaucher disease, type iii
https://www.malacards.org/card/gaucher_disease_type_iii
- Mount Sinai School of Medicine
<https://icahn.mssm.edu/research/gaucher/what-is>

- Orphanet: Gaucher disease
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=355
- Orphanet: Gaucher disease-ophthalmoplegia-cardiovascular calcification syndrome
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2072

Patient Support and Advocacy Resources

- Canadian MPS Society
<http://www.mpssociety.ca/>
- National Gaucher Foundation
<https://www.gaucherdisease.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/gaucher-disease/>
- National Tay Sachs and Allied Diseases Association
<https://www.ntsad.org/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/gaucher.html>

Clinical Information from GeneReviews

- Gaucher Disease
<https://www.ncbi.nlm.nih.gov/books/NBK1269>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Gaucher+Disease%5BMAJR%5D%29+AND+%28Gaucher+disease%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- GAUCHER DISEASE, TYPE I
<http://omim.org/entry/230800>
- GAUCHER DISEASE, TYPE II
<http://omim.org/entry/230900>
- GAUCHER DISEASE, TYPE III
<http://omim.org/entry/231000>
- GAUCHER DISEASE, TYPE IIIC
<http://omim.org/entry/231005>

Medical Genetics Database from MedGen

- Gaucher disease
<https://www.ncbi.nlm.nih.gov/medgen/42164>

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