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.

Genetic Changes

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
- kerasin histiocytosis
- kerasin lipoidosis
- kerasin thesaurismosis
- lipoid histiocytosis (kerasin type)

Diagnosis & Management

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/

Other Diagnosis and Management Resources

- Baby's First Test
  http://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/

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

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

- Disease InfoSearch: Gaucher Disease
  http://www.diseaseinfosearch.org/Gaucher+Disease/3000
- JAMA Patient Page: Gaucher disease
  https://jamanetwork.com/journals/jama/fullarticle/208870
- MalaCards: gaucher disease, type i
  http://www.malacards.org/card/gaucher_disease_type_i
- MalaCards: gaucher disease, type ii
  http://www.malacards.org/card/gaucher_disease_type_ii
- MalaCards: gaucher disease, type iii
  http://www.malacards.org/card/gaucher_disease_type_iii
- Mount Sinai School of Medicine
  http://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

GeneReviews

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

ClinicalTrials.gov

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

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

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

MedGen

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

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4141347/

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