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URL of this page: <https://medlineplus.gov/gaucherdisease.html>

Gaucher Disease

Gaucher disease is a rare, inherited disorder. It is a type of **lipid metabolism disorder**

[<https://medlineplus.gov/lipidmetabolismdisorders.html>] . If you have it, you do not have enough of an enzyme called glucocerebrosidase. This causes too much of a fatty substance to build up in your spleen, liver, lungs, bones and, sometimes, your brain. This prevents these organs from working properly.



There are three types:

- Type 1, the most common form, causes liver and spleen enlargement, bone pain and fractures [<https://medlineplus.gov/fractures.html>] (broken bones), and, sometimes, lung [<https://medlineplus.gov/lungdiseases.html>] and kidney [<https://medlineplus.gov/kidneydiseases.html>] problems. It does not affect the brain. It can occur at any age.
- Type 2, which causes severe brain damage, appears in infants. Most children who have it die by age 2.
- In type 3, there may be liver and spleen enlargement. The brain is gradually affected. It usually starts in childhood or adolescence.


Gaucher disease has no cure. Treatment options for types 1 and 3 include medicine and enzyme replacement therapy, which is usually very effective. There is no good treatment for the brain damage of types 2 and 3.

NIH: National Institute of Neurological Disorders and Stroke


Learn More

- Gaucher Disease and Pregnancy [<https://mothertobaby.org/fact-sheets/gaucher-disease-pregnancy/>]
(Organization of Teratology Information Specialists)
Also in Spanish [<https://mothertobaby.org/es/hojas-informativas/la-enfermedad-de-gaucher/>]
- Learning about Gaucher Disease [<https://www.genome.gov/Genetic-Disorders/Gaucher-Disease>]
 (National Human Genome Research Institute)
- Lipid Storage Diseases [<https://www.ninds.nih.gov/health-information/disorders/lipid-storage-diseases>]
 (National Institute of Neurological Disorders and Stroke)

Genetics

- Gaucher disease: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/gaucher-disease>]
 (National Library of Medicine)

Clinical Trials

- ClinicalTrials.gov: Gaucher Disease [<https://clinicaltrials.gov/search?cond=%22Gaucher+Disease%22&aggFilters=status:not%20rec>]  (National Institutes of Health)

Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: Age-related inflammatory biomarkers in early-onset osteoporosis in females with Gaucher disease. [https://www.ncbi.nlm.nih.gov/pubmed/40671914]
- Article: Expression Profiles of Exosomal miRNAs in Gaucher Patients and Their Association... [https://www.ncbi.nlm.nih.gov/pubmed/40619747]
- Article: The clinical utility of neurofilament light chain for early detection and... [https://www.ncbi.nlm.nih.gov/pubmed/40570595]
- Gaucher Disease -- see more articles [https://pubmed.ncbi.nlm.nih.gov/?term=%22Gaucher+Disease%22%5Bmajr%3Anoexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bedat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+]

Patient Handouts

- Gaucher disease [https://medlineplus.gov/ency/article/000564.htm] (Medical Encyclopedia)
Also in Spanish [https://medlineplus.gov/spanish/ency/article/000564.htm]



National Institutes of Health

The primary NIH organization for research on *Gaucher Disease* is the National Institute of Neurological Disorders and Stroke [http://www.ninds.nih.gov/]

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