



## Gaucher Disease

John L. Zeller; Alison E. Burke; Richard M. Glass

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# Gaucher Disease

**G**aucher disease is a chronic, progressive, inherited disorder named for the French physician who first described it. It is caused by a deficiency of a specific enzyme called **glucocerebrosidase**. This enzyme breaks down and recycles certain **lipids** (fats). In people with Gaucher disease, these lipids, **glucocerebrosides**, accumulate within cells and interfere with the normal functioning of various organs (liver, spleen, lungs, bone marrow, and brain). The disease is classified into 3 types. In type 1, there is no brain involvement. It is found in adults, accounts for 95% of all cases, and can sometimes have no symptoms. The type 2 condition is uncommon and severe. Brain damage can be extensive. Children usually die by 2 years of age. Type 3 occurs in children and adolescents. It tends to be chronic and slowly progressive and the central nervous system is affected.

It is estimated that approximately 1 in 40 000 to 60 000 persons in the general population has Gaucher disease, or about 10 000 people worldwide. It is the most common disorder in a group of more than 40 diseases classified as **lysosomal storage disorders** (LSDs). Grouped together, LSDs affect 1 in every 7700 babies born. The September 19, 2007, issue of *JAMA* includes an article about genetic screening and treatment for Gaucher disease.

## RISK FACTORS

Gaucher disease may occur among every nationality and ethnic group but it is most common among **Ashkenazi Jews** (individuals of particular Jewish descent from Eastern or Central Europe). In this particular ethnic group, the incidence is 1 in 450 persons.

## TESTING AND TREATMENT

A blood test to measure glucocerebrosidase enzyme activity is currently used to determine whether a person has Gaucher disease or may be a carrier. With a confirmation of the diagnosis, disease management has shifted to disease-specific therapies:

- **Enzyme replacement therapy (ERT)**—The goal of ERT is to provide an appropriate amount of needed enzyme.
- **Substrate reduction therapy (SRT)**—The goal is to minimize the amount of production and accumulation of waste material within cells.
- **Symptom management**—Treatment includes pain reduction therapies, blood transfusions, orthopedic surgery, and possible **splenectomy** (removal of the spleen).
- **Psychological care**—Professional counseling can help patients better manage the difficulties of their disease and the lifestyle changes that might be required.

## SIGNS AND SYMPTOMS

Symptoms of Gaucher disease can vary from the very mild, or even none, to severe. Bone pain or fracture is often the first symptom. Other symptoms may include

- Skeletal abnormalities
- **Hepatomegaly** (enlarged liver)
- **Splenomegaly** (enlarged spleen)
- Anemia (reduced number of red blood cells)
- Excessive fatigue
- Bleeding and easy bruising due to thrombocytopenia (low platelet count)
- Mental retardation
- Dementia
- **Pingueculae** (yellow spots in the eyes)
- Abnormal eye movements

## FOR MORE INFORMATION

- National Gaucher Foundation  
www.gaucherdisease.org
- National Organization for Rare Disorders (NORD)  
www.rarediseases.org

## INFORM YOURSELF

To find this and previous JAMA Patient Pages, go to the Patient Page link on JAMA's web site at [www.jama.com](http://www.jama.com). Many are available in English and Spanish.

Sources: National Gaucher Foundation, National Organization for Rare Disorders

John L. Zeller, MD, PhD, Writer

Alison E. Burke, MA, Illustrator

Richard M. Glass, MD, Editor

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