What is Gaucher disease?

An overview

Gaucher (pronounced go-SHAY) disease is a rare genetic disorder. It is thought to affect 1 in 50,000 to 1 in 100,000 people worldwide. A higher frequency of the disease is seen in people of Eastern and Central European (Ashkenazi) Jewish heritage. In this group, Gaucher disease affects about 1 in 600 people.

In a person with Gaucher disease, cells do not produce enough of an enzyme called glucocerebrosidase (pronounced GLOO-ko-SER-e-bro-sy-daze). This enzyme is needed to break down a fatty substance called glucocerebroside (pronounced GLOO-ko-SER-e-bro-side). Without enough of this enzyme, the fatty substance builds up in some cells. These cells are called Gaucher cells.

Over time, Gaucher cells collect in various organs, causing the signs and symptoms of Gaucher disease.

Type 1 Gaucher disease

Type 1 is the most common form of Gaucher disease. It accounts for more than 90% of all cases. It is the only type of Gaucher disease that is considered “nonneuronopathic,” meaning it does not involve the central nervous system like rarer forms of the disease.

Signs and symptoms

Signs and symptoms of Type 1 Gaucher disease include:

- Increased spleen size
- Increased liver size
- Low hemoglobin level
- Low platelet count

How is Gaucher disease inherited?

Gaucher disease and genetics

Gaucher disease is inherited, meaning it is passed down from one generation to the next through our genes. A pair of genes determines each of our traits—1 inherited from each parent. Gaucher disease can only develop when a person inherits 2 abnormal copies of the gene responsible for producing the enzyme glucocerebrosidase. If a person inherits 1 abnormal copy of this gene, he or she does not have Gaucher disease, but is considered a carrier. A carrier could pass the gene that could cause the disease on to his or her children.

Raising awareness of Gaucher disease together

Gaucher disease is a rare, inherited condition:

Type 1 is the most common form of Gaucher disease. It accounts for more than 90% of all cases.

- Type 1 Gaucher disease affects 1 in 50,000 to 1 in 100,000 people
- Among those of Ashkenazi Jewish heritage, it affects as many as 1 in 600 people

If Gaucher disease runs in your family, or a family member is a carrier, consider getting tested for the condition.

Worldwide prevalence

1 in 50,000 people to **1 in 100,000 people**

Among those of Ashkenazi Jewish heritage

1 in 600 people

How Gaucher disease is diagnosed

Diagnosing Gaucher disease can sometimes be a challenge. Some people may have symptoms for many years without knowing they have the disease. When symptoms develop, they can often be confused with other diseases. For these reasons, it can sometimes take time before a diagnosis is made.

Genetic testing can be used to identify if a person is a carrier for Gaucher disease. The diagnosis can be confirmed through enzyme activity testing.

Enzyme testing

Enzyme testing measures glucocerebrosidase activity in the blood and urine. Glucocerebrosidase is the enzyme that is not working correctly in a person with Gaucher disease.

Gaucher disease is a progressive disease. Left untreated, it can become debilitating. That is why an early diagnosis is important. Although there is currently no cure for Gaucher disease, treatment can help people manage some of the symptoms of the disease. If you or a loved one have been diagnosed with Gaucher disease, talk to your doctor about possible treatment options.

Commitment to the Gaucher community

Pfizer—committed to discovering new therapies for rare diseases

Rare diseases are among some of the most challenging illnesses and collectively affect millions of patients worldwide, representing an opportunity to apply our knowledge and expertise to help make a significant impact in addressing unmet medical needs. The Pfizer focus on rare diseases builds on more than a decade of experience and a global portfolio of over 20 medicines approved worldwide that treat rare diseases in the areas of hematology, neuroscience, inherited metabolic disorders, pulmonology, and oncology.

Pfizer Patient Affairs Liaison

Patient Affairs Liaisons are available to help provide support for people with Type 1 Gaucher disease, as well as for their parents and caregivers. Patient Affairs Liaison offerings include:

- Answering general, nonmedical questions about treatment and about Pfizer
- Coordinating Pfizer's educational offerings for people with Type 1 Gaucher disease and their parents or caregivers
- Accepting feedback about offerings provided by the Pfizer Gaucher Personal Support (GPS) program
- Providing assistance in finding additional Gaucher disease–related resources such as advocacy groups and community programs

A Pfizer Patient Affairs Liaison can be reached at **PfizerPAL.com**.

Pfizer Rare Disease