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What are the Gaucher Disease Symptoms

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Q: What is Gaucher disease?

A: Gaucher disease is a rare, inherited disease. It was first described by Dr. Philippe Gaucher in 1882 and is caused by genetic mutations (a permanent change in the DNA of a gene) received from both parents. In people with Gaucher disease, the body's cells do not produce enough of an enzyme called glucocerebrosidase (pronounced "GLOO-ko-SERe-bro-sy-dase"). Without enough of this enzyme, people with Gaucher disease can't break down a fatty substance found in cells known as glucocerebroside (pronounced "GLOO-ko-SER-e-bro-side"). This fatty substance builds up in some cells and enlarges them. These enlarged cells are called Gaucher cells.

Over time, Gaucher cells collect in some organs including the liver, spleen, lungs, and bone marrow, damaging them until they can't work the way they should. The bone-related damage can be especially painful and may eventually limit a person's mobility. In rare cases, Gaucher cells can also collect in the brain and result in a more severe form of the disease.

Q: Who gets Gaucher disease and how do they get it?

A: Gaucher disease is an inherited condition, meaning it's passed down from one generation to the next through the genes of one's parents. As with all of our traits, we inherit a pair of genes (one gene from each parent) that is responsible for our ability to produce glucocerebrosidase. A person gets Gaucher disease when the mother and father each carry an abnormal copy of this gene and the child receives an abnormal copy from both parents. If a person inherits only one of these abnormal genes, they are considered a carrier. That means they do not have the disease but can pass the gene on to their children.

Gaucher disease can affect people of any ethnicity, but it is more common in people of Ashkenazi Jewish background.

Q: How rare is Gaucher disease?

A: About 1 in 57,000 people have Gaucher disease or approximately 125,000 people worldwide. About 1 in 855 people of Ashkenazi Jewish descent have Gaucher disease.

Q: What are the signs and symptoms of Gaucher disease?

A: The signs and symptoms of Gaucher disease vary from one person to another. Some people have very mild symptoms and live with them for years before being diagnosed with the disease. Others have more severe symptoms.

Common signs and symptoms include:

- Low hemoglobin (anemia)
- Low platelet count
- Enlarged liver
- Enlarged spleen
- Bone pain
- Bone disease
- Fatigue
- Easy bruising

Less common signs and symptoms include:

- Heart and lung problems
- Cognitive impairment
- Seizure
- Dementia

Q: What are the different types of Gaucher disease?

A: There are three different types of Gaucher disease.

Type 1 Gaucher disease is the most common and accounts for more than 90% of cases. The signs and symptoms of type 1 can begin at any age and they usually include anemia bruising, bleeding, abdominal pain (caused by an increase in spleen and liver size), bone pain and growth problems. People with type 1 Gaucher disease can often expect to have a nomal life span. Type 1 is also sometimes called the adult form of Gaucher disease, although the cause is present from the time of conception. Type 1 Gaucher disease occurs worldwide in all populations, but is most prevalent in the **Ashkenazi Jewish population.**

Type 2 Gaucher disease is characterized by abnormalities of the central nervous system and is usually fatal during the first two to four years of life. Type 2 Gaucher disease equally affects people worldwide but is very rare.

Type 3 Gaucher disease is also very rare and equally common among ethnic populations. In this type of Gaucher disease, the neurologic symptoms progress slowly. Symptoms usually develop in childhood and continue through adulthood.

Treatment can help with some of the symptoms of the disease, especially for people who receive a diagnosis and begin treatment early.

Q: How is Gaucher disease diagnosed?

A: The most common symptoms of Gaucher disease may cause a doctor to suspect the condition, but these symptoms cannot be used on their own as a way to diagnose the disease. Instead, a diagnosis of Gaucher disease can be made with a blood test. The test will measure glucosylceramidase enzyme activity in the blood.

For people with a known family history of Gaucher disease, a diagnosis can be confirmed with genetic testing. Carrier testing can also be done for individuals who have a family history. If the child's mother and father are both carriers, there is a 25% chance that the child will be affected by Gaucher disease and a 50% chance that the child will be a carrier.

Genetic counseling is recommended for prospective parents with a family history of Gaucher disease, and prenatal testing can also be done to learn if a fetus in the womb has Gaucher disease. Efforts are also underway to include Gaucher disease testing as part of Newborn Screenings for all infants born in the United States.

Q: Can Gaucher disease be treated?

A: Although there is currently no cure for Gaucher disease, treatment can help people manage some of the symptoms of the disease. Enzyme replacement therapy can be used in most people with types 1 and 3 Gaucher disease. There are no effective treatments for those with types 2 and 3 Gaucher disease and severe brain involvement. It is best to discuss treatment options with a treating physician.

Q. Where can people with Gaucher disease and their family members find support for coping with the condition?

A. There are several organizations dedicated to providing emotional and financial support for people affected by Gaucher disease. They include the National Gaucher Foundation the National Organization for Rare Disorders (NORD), Genetic Alliance, and the Genetic and Rare Diseases Information Center (GARD).

Sukriti Bagal, MD was the Medical Director of Rare Diseases and Clinical Development at Pfizer.

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References

- **1.** Meikle PJ, Hopwoode JJ, Clague AE, Carey WF. Prevalence of lysosomal storage disorders. *JAMA*. 1999 Jan 20; 281(3): 249-254. doi: 10.1001/jama.281.3.249
- **2.** Pastores GM and Hughes DA. Gaucher disease. National Institutes of Health Web site. Accessed January 9, 2014.

- **3.** United States Census Bureau. U.S. census bureau current population. Accessed January 9, 2014.
- **4.** Jmoudiak M and Futerman AH. Gaucher disease: pathological mechanisms and modern management. *Br J Haematol*. 2005; 129: 178-188. doi: 10.1111/j.1365-2141.2004.05351.x
- **5.** Grabowski GA. Gaucher disease and other storage disorders. *Hematology Am Soc Hematol Educ Program*. 2012; 1: 13-18. doi: 10.1182/asheducation-2012.1.13
- **6.** Genetics Home Reference. What is a gene mutation and how do mutations occur? Accessed January 9, 2014.
- **7.** Merriam-Webster. Glucocerebrosidase. Accessed January 14, 2016.
- **8.** Merriam-Webster. Glucocerebroside. Accessed January 9, 2016.
- **9.** Pastores GM, Weinreb NJ, Aerts H, Andria G et al. Therapeutic goals in the treatment of Gaucher disease. *Semin Hematol*. 2004; 41(suppl 5): 4-14. doi: 10.1016/j.seminhematol.2004.07.009
- **10.** Rosenbloom BE, Weinreb NJ, Zimran A, Kacena KA et al. Gaucher disease and cancer incidence: a study from the Gaucher registry. *Blood*. 2005 Jun 15; 105(12): 4569-4572. doi: 10.1182/blood-2004-12-4672
- **11.** Zimran A. How I treat Gaucher disease. *Blood*. 2011 Aug 11; 118(6): 1463-1471. doi: 10.1182/blood-2011-04-308890
- **12.** Grabowski GA. Phenotype, diagnosis, and treatment of Gaucher's disease. *Lancet*. 2008; 372: 1263-71. doi: 10.1016/S0140-6736(08)61522-6
- **13.** Kaplan P, Baris H, De Meirleir L, Di Rocco M et al. Revised recommendations for the management of Gaucher disease in children. *Eur J Pediatr*. 2013 Apr; 172(4): 447-58. doi: 10.1007/s00431-012-1771-z
- **14.** Marsden D and Levy H. Newborn screening of lysosomal storage disorders. *Clin Chem*. 2010; 56(7): 1071-1079. doi: 10.1373/clinchem.2009.141622

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