

Gaucher disease and the family

An information brochure
for people affected by
Gaucher disease

Imma (person with Gaucher disease type 3)

Gaucher disease is a rare, inherited condition. The aim of this brochure is to answer some of the questions you may have about Gaucher disease – its cause, symptoms, diagnosis, inheritance and treatment.





Gustavo (person with Gaucher disease type 1)



Katrin (person with Gaucher disease type 1)

What is Gaucher disease?

Gaucher disease is a rare inherited disease. It is caused by a genetic mutation that prevents an enzyme – called **glucocerebrosidase** – from working as it should.¹ It is this enzyme's job to break down a substance called **glucocerebroside** (also called glucosylceramide). If the enzyme doesn't work properly or doesn't work at all, then the glucocerebroside can build up in cells over time. This can cause problems in the body.²

What are the symptoms of Gaucher disease?

The symptoms of Gaucher disease vary from person to person.³ In some people, Gaucher disease can be mild or may have no symptoms at all.^{3,4}

Symptoms experienced by some people with Gaucher disease*

Most common symptoms

Swollen or tender stomach or abdomen due to an enlarged spleen (known as splenomegaly)⁴⁻⁶

Persistent unexplained bleeding or frequent bruising (caused by a low platelet count, which is known as thrombocytopenia)⁴⁻⁶



Other common symptoms

Slow growth (in children)^{5,6}

Liver can also be enlarged (known as hepatomegaly)⁴

Bone/joint pain or bone fractures⁴⁻⁶

*These symptoms are not specific to Gaucher disease and might not constitute a diagnosis of Gaucher disease. Please consult your doctor if concerned.

Elaine: *“By the age of about 4 or 5, I was showing the classic signs of Gaucher disease: a very pronounced tummy, very pale skin, my limbs were very thin and I would bruise very easily.”*

How is Gaucher disease diagnosed?

The symptoms of Gaucher disease vary a lot and can resemble those of many other, more common conditions.⁵ This means that Gaucher disease is not always easy for doctors to diagnose, and the process of receiving a diagnosis can be slow for some patients.⁵ However, if your doctor thinks you might have Gaucher disease, then the diagnosis can be confirmed with a simple blood test.

Ivana: *“Doctors from my city hospital, they did not know what was happening. They hadn't even heard of Gaucher disease.”*

How is Gaucher disease inherited?

Gaucher disease is a hereditary condition – it is passed down from parents to their children.

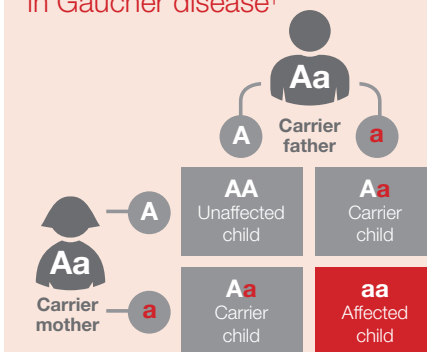
Every cell in the human body contains chromosomes – thread-like structures carrying genetic information – that exist as pairs. One chromosome in each pair is inherited from the mother, and the other from the father. For each gene, a person therefore inherits one copy, or **allele**, from each parent.⁷ The gene responsible for Gaucher disease is found on chromosome 1.¹

To develop Gaucher disease, the person must inherit two copies of a mutated Gaucher disease gene – one from each parent. This is called **autosomal recessive** inheritance.⁷

People who inherit just one chromosome containing a mutated Gaucher disease gene are called **carriers**. They cannot develop Gaucher disease themselves, but they can pass the mutated gene on to their children.⁷

¹If both parents are carriers – each having one Gaucher disease mutated gene (**a**) and one normal gene (**A**) – there is a 1 in 4 chance that their child will develop Gaucher disease. Also, there is a 2 in 4 chance that the child will be a carrier, and a 1 in 4 chance that the child will be unaffected. These odds are the same for each pregnancy. This means that already having three children without Gaucher disease does not mean that the fourth child will definitely develop it.⁷

Autosomal recessive inheritance in Gaucher disease[†]



Virginia: *“I think Gaucher disease as a whole has made my family more close together. We have connected in a way that, I don't know if other families do.”*



Ivana (person with Gaucher disease type 1)

How is Gaucher disease treated?

There is no cure for Gaucher disease, but certain treatments are available to help with the management of the disease. These are not appropriate for everyone who has Gaucher disease. Your doctor can discuss suitable management options with you.

Virginia: *“I find myself very fortunate to have been diagnosed at such an early age, and also being able to get my treatment right away. Because now I feel really healthy and live a full life.”*

Support services



International Gaucher Alliance (IGA)

An organization representing the interests of Gaucher Patients and Groups throughout the world

gaucheralliance.org

References

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