









Gaucher disease and inheritance

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 **glucosylceramide**. If the enzyme doesn’t work properly or doesn’t work at all, then the glucosylceramide can build up in cells over time. This can cause problems in the body.²

How does Gaucher disease run in the family?

Gaucher disease follows **autosomal recessive inheritance**. This means that, to develop Gaucher disease, a person must inherit two copies (alleles) of a mutated Gaucher disease gene – one from each parent. Therefore, the chances of a child having Gaucher disease depends on the parents’ mutation status. (Someone who has one mutated Gaucher disease gene and one normal gene is a **carrier**; they cannot develop Gaucher disease, but can pass the mutated gene on to their children.)³

Parents’ mutations	Risk of:		
	being unaffected	being a carrier	having Gaucher disease
 Carrier  Carrier	25% (1 in 4)	50% (2 in 4)	25% (1 in 4)
 Gaucher disease  Non-affected		100% (4 in 4)	
 Gaucher disease  Gaucher disease			100% (4 in 4)

The gene responsible for Gaucher disease can be passed on for several generations, potentially affecting close and distant relatives.

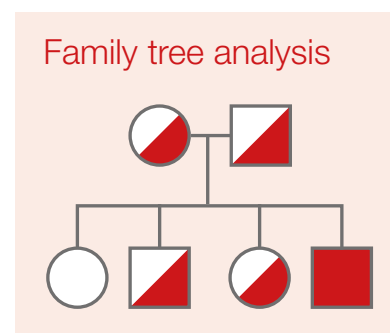
Elaine: *“It’s of great value having a brother to talk to that understands totally what I’m going through.”*

What is family tree analysis?

To work out the risk of inheriting Gaucher disease for relatives, a doctor will ask for the family's medical history, and map out a family tree using symbols to represent genetic relationships; this is called family tree analysis – also called pedigree analysis.

In a family tree (see diagram), squares represent males and circles represent females. The fully shaded symbols represent individuals who have two alleles of the mutated gene, and therefore have Gaucher disease. The half-shaded symbols represent carriers of the mutated gene.

This simple method allows a large amount of information to be condensed into a straightforward diagram, which doctors can then use to see inheritance patterns and calculate the risk for each family member.



What should I do if I or a member of my family has been diagnosed with Gaucher disease?

Gaucher disease is a complex condition with varying symptoms that can be missed or wrongly diagnosed. Accurate diagnosis means that help can be accessed in a timely manner from the appropriate specialists.

The hereditary nature of Gaucher disease means that a diagnosis not only has important health consequences for the person, but may also have implications for their family. It is therefore important to speak to a doctor or a genetic counsellor about drawing a family tree. While not a diagnostic tool, family tree analysis can help to work out if any relatives should be tested.

How to discuss Gaucher disease with family members

Telling family members that they might be at risk of Gaucher disease can seem like a daunting task. However, there is plenty of help on offer from doctors, genetic nurses, or genetic counsellors on how best to communicate with relatives. Do seek the advice of these medical professionals before speaking to your family, so that you are prepared for a frank and considered discussion, which will facilitate the early diagnosis and treatment of those at risk.

There are also a number of patient organisations that can provide further information and support.

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

References

1. Burrow TA, *et al.* Prevalence and management of Gaucher disease. *Pediatr Health Med Ther* 2011;2:59–73.
2. Mistry PK, *et al.* A reappraisal of Gaucher disease diagnosis and disease management algorithms. *Am J Hematol* 2011;86:110–115.
3. Gauchers Association. Inheritance. Available at: http://gaucher.org.uk/about_gaucher/inheritance; Accessed: August 2018.

