

Gaucher disease and your family

Information leaflet for patients with Gaucher disease



This leaflet hopes to answer some of the questions you may have about the inheritance of Gaucher disease, and offers advice and support in communicating implications of this diagnosis to your immediate and extended family.

References

1. Mistry, PK. et al. Consensus conference: A reappraisal of Gaucher disease - diagnosis and disease management algorithms. *Am J Hematol.* 2011; 86(1): 110-115
2. Gaucher Association. Inheritance. Available at: http://www.gaucher.org.uk/about_gaucher/inheritance [Accessed January 2015]
3. Burrows, TA. et al. Prevalence and management of Gaucher disease. *Pediatric Health, Medicine and Therapeutics.* 2011; 2: 59-73

What is Gaucher disease?

Gaucher disease is a rare inherited disease caused by the deficiency, absence or incomplete functioning of an enzyme called glucocerebrosidase.¹

Over time, this can result in the accumulation of a waste substance called glucocerebroside in cells.¹

How does Gaucher disease run in the family?

Gaucher disease is not contagious, but it is a hereditary condition that can be passed down from parents to their children in the pattern illustrated below.²

Every cell in the human body contains chromosomes, which are thread-like structures carrying genetic information. These chromosomes exist in pairs, and each person will inherit one of each pair from their mother, and the other from their father.²

The gene responsible for Gaucher disease is found on chromosome 1.³ It is necessary to inherit two copies of this chromosome containing a mutated version of this Gaucher disease gene (one from each parent) in order to develop the disease. Gaucher disease is therefore known as an *autosomal recessive* disease.²

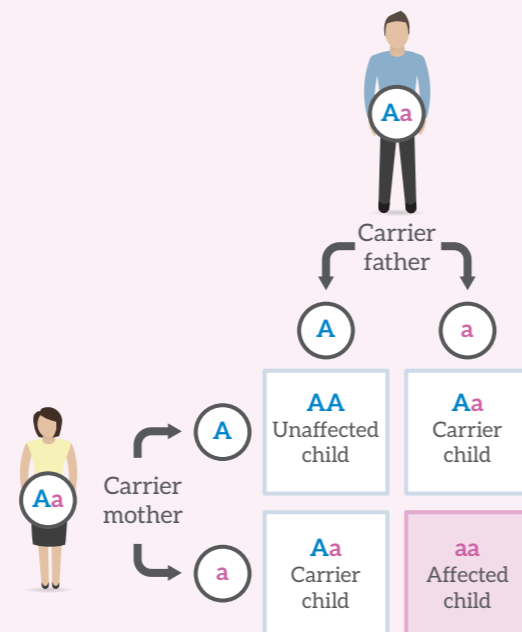
A person that has one chromosome containing a mutated Gaucher disease gene and one chromosome containing a normal gene will **not** develop Gaucher disease. This type of person is called a *carrier*.²

The way in which Gaucher disease can be passed on in families is shown in the diagram on the right.

If both parents are carriers of Gaucher disease (each parent has one Gaucher disease mutated gene, 'a', and one normal gene, 'A') there is a risk that:²

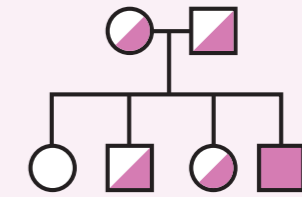
- 1 in 4 (25%) of their children will inherit two normal copies of the gene (A), and will therefore be unaffected by Gaucher disease.
- 2 in 4 (50%) of their children will inherit only one copy of the mutated gene, and will therefore be carriers.
- 1 in 4 (25%) of their children will inherit two copies of the mutated Gaucher disease gene (a), and will therefore develop Gaucher disease.

Autosomal recessive inheritance



The gene responsible for Gaucher disease can be passed on for several generations, thereby potentially affecting many close and distant relatives. To work out the risk of inheriting Gaucher disease for your relatives, a doctor will ask for your family's medical history, and map out a family tree using symbols to represent genetic relationships; this is called *pedigree analysis*.

The family tree diagram shown here is in fact the same as the diagram showing the inheritance of an autosomal recessive trait, but is expressed in a much simpler form.



What is pedigree analysis?

In a pedigree family tree, squares represent males, circles females, the shaded symbols are individuals who have the mutated gene. 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 have been diagnosed with Gaucher disease?

Gaucher disease is a complex condition with varying symptoms that can be missed or wrongly diagnosed. As such, an accurate diagnosis means that you can get help in a timely manner from the appropriate specialists.

Due to the hereditary nature of this disorder, a diagnosis not only has important health

consequences for you, but may also have implications for your family. It is therefore important that you speak to your doctor or a genetic counsellor about drawing a pedigree family tree. While not a diagnostic tool, pedigree analysis can help to work out if any of your relatives should be tested.

How should I tell my family about my condition?

Telling your 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 your doctor, genetic nurse, or genetic counsellor on how best to communicate with your relatives. Do seek the advice of these medical professionals before you speak 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. Their details may be found at the back of this leaflet.