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
What is Gaucher disease?

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

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

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.

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.