

GAUCHER
DISEASE
EARLY
DIAGNOSIS

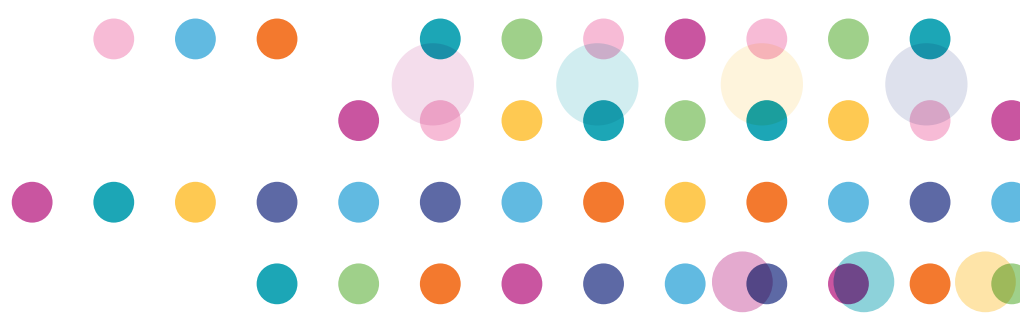
Prepared and funded by Shire.

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GAUCHER DISEASE AND YOUR FAMILY

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**An information brochure for
patients with Gaucher disease**



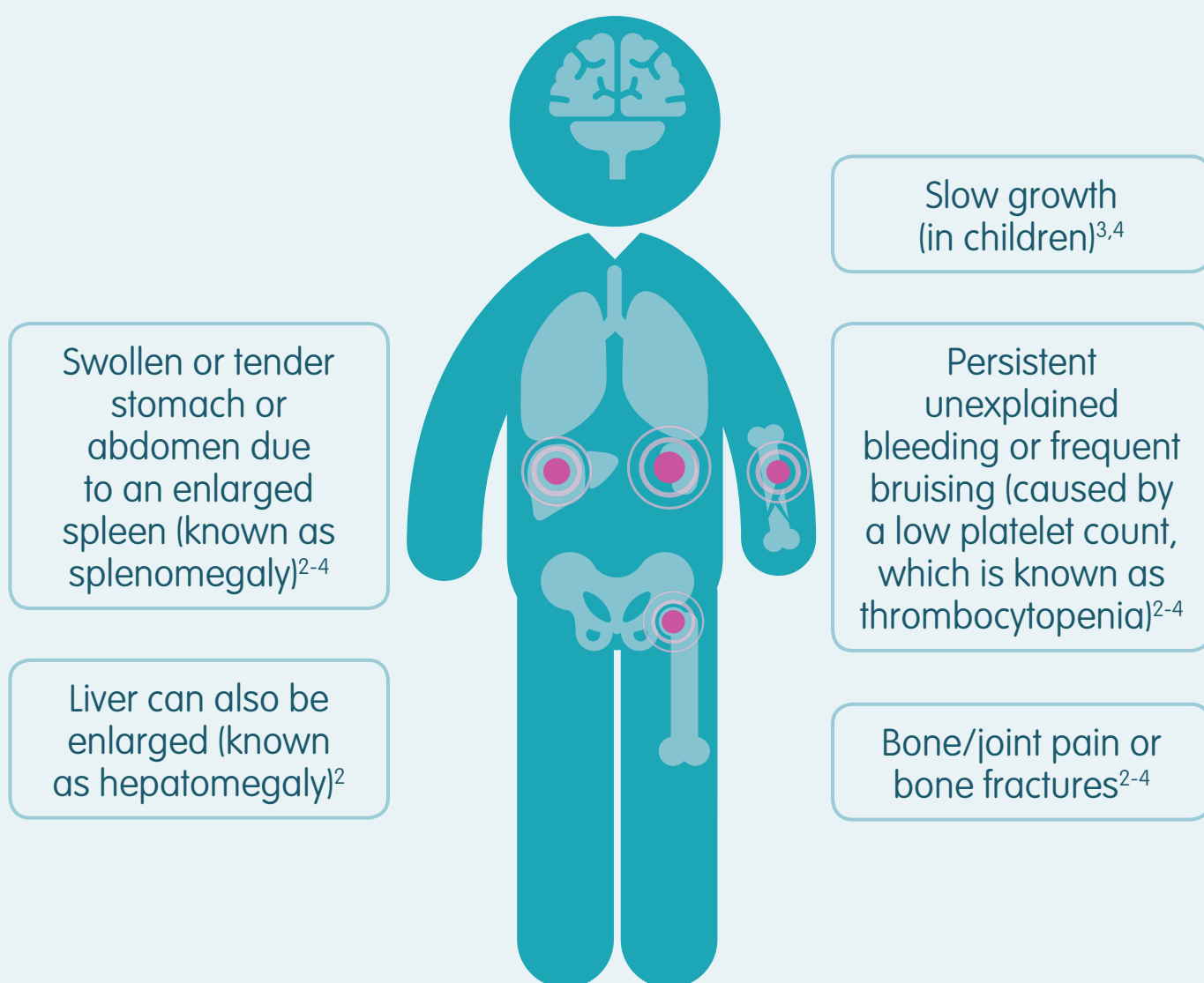
Gaucher disease is a rare, inherited condition. This brochure hopes to answer some of the questions you may have about the causes, signs and symptoms of Gaucher disease and how it is inherited. It also offers recommendations on what to do if you or a member of your family has been diagnosed with the condition.

What are the symptoms of Gaucher disease?

The symptoms of Gaucher disease vary from person to person.¹ In some people, Gaucher disease can be mild or may have no symptoms at all.^{1,2} The most common symptoms are:

- A swollen or tender stomach or abdomen²⁻⁴
- Persistent bleeding or frequent bruising²⁻⁴

The symptoms that some people with Gaucher disease can experience*



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

What causes Gaucher disease?

Gaucher disease is a rare inherited disease caused by the deficiency, absence or incomplete functioning of an enzyme called glucocerebrosidase.⁵ Over time, this can result in the accumulation of a waste substance called glucosylceramide in cells.⁶

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

How is Gaucher disease diagnosed?

The symptoms of Gaucher disease vary a lot and can resemble many other, more common, conditions.³ 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.³ However, if your doctor suspects you might have Gaucher disease then the diagnosis can be confirmed with a simple blood test.

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. Every cell in the human body contains chromosomes, which are thread-like structures carrying genetic information. Chromosomes exist in pairs. For each pair, one chromosome is inherited from the mother, and the other from the father.

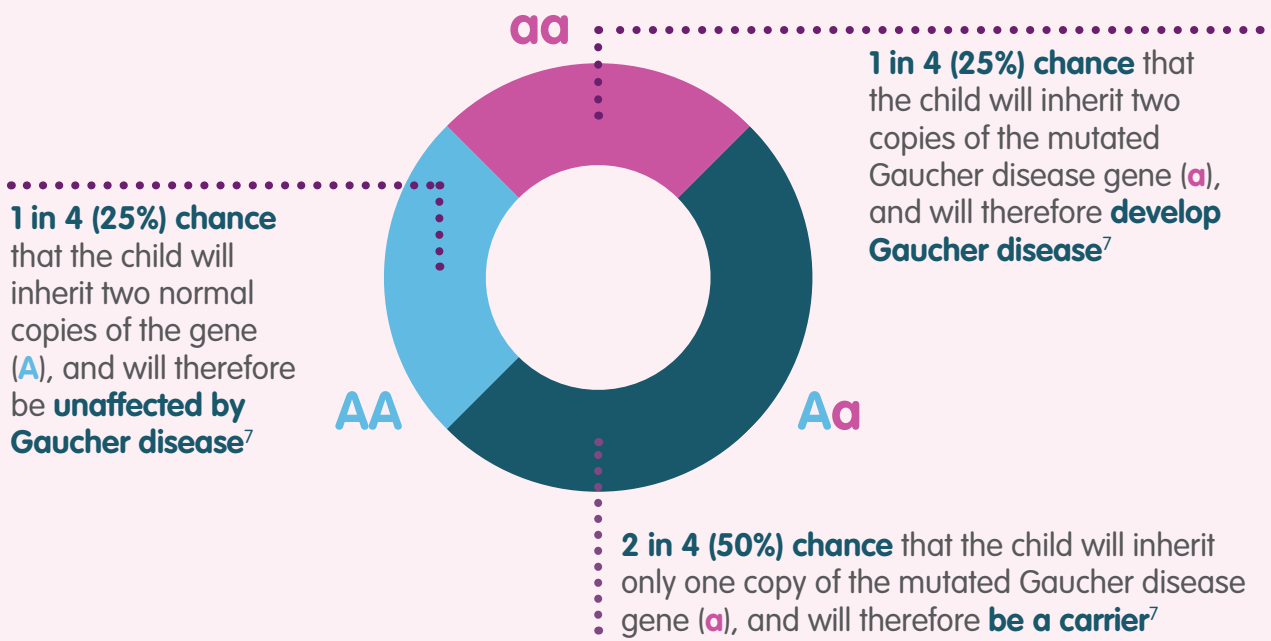
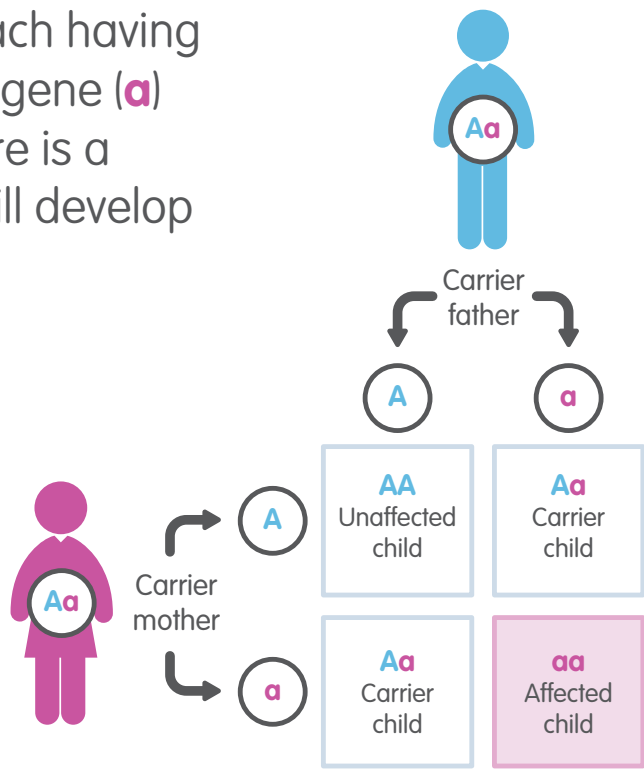
The gene responsible for Gaucher disease is found on chromosome 1.⁵ To develop Gaucher disease, the person must inherit two copies of chromosome 1 containing a mutated version of the Gaucher disease gene (one from each parent).⁷ Gaucher disease is, therefore, known as an autosomal recessive disease.⁷

Other people can be carriers of Gaucher disease if they have one chromosome containing a mutated Gaucher disease gene and one chromosome containing a normal gene.⁷ Carriers cannot develop Gaucher disease themselves, but they can pass the mutated gene on to their children.⁷



Autosomal recessive inheritance

If 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.⁷

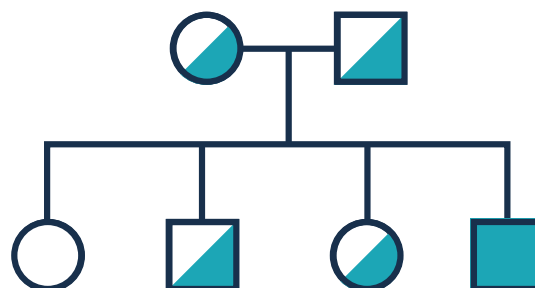


These odds are the same for each pregnancy.⁷ This means that already having three children without Gaucher disease does not mean that the fourth child will definitely develop it.

Even if a child inherits two copies of the mutated Gaucher disease gene, the severity of symptoms can vary considerably, with some experiencing no obvious Gaucher disease symptoms.¹

What should I do if I have been diagnosed with Gaucher disease?

An accurate diagnosis means that you can get timely help from the appropriate specialists. Due to the hereditary nature of Gaucher disease, 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 counselor about drawing a pedigree family tree.



While it is not a diagnostic tool, pedigree analysis can help to work out if any of your relatives should be tested. Your doctor will construct a pedigree family tree, with squares representing males and circles representing females. Symbols that are shaded are individuals who have the mutated gene, whilst 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 that helps the doctor to see the inheritance pattern.

How should I tell my family about my condition?

Telling your family members that they might be at risk of Gaucher disease can seem daunting. However, there is plenty of help on offer from your doctor, genetic nurse, or genetic counselor on how best to talk to 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 help get an early diagnosis and treatment for those at risk. There are also a number of patient organizations that can provide further information and support.

Is there a treatment for Gaucher disease?

There is no cure for Gaucher disease, however, certain treatments are available to help with the management of the disease, if appropriate for you - your doctor can discuss this with you.



Support services

[To be completed by individual countries with details of local services, patient societies etc]

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References

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