What is Gaucher?

A guide for people living with type 1 Gaucher disease

Takeda

beMi®

Inside this booklet

This booklet provides general information about Gaucher. It is not intended to replace advice from a healthcare professional. Readers are advised to discuss medical questions with their healthcare team.

What is Gaucher?

- Gaucher (pronounced ‘go-shay’) disease is a type of rare genetic condition called a lysosomal storage disorder. It is something you inherit from your parents.

Gaucher is caused by changes in a certain gene. These changes mean that an enzyme called glucocerebrosidase does not work as it should.

- The enzyme helps break down a type of fat (glucocerebroside) that builds up in the cells in the body.
- Without enough of the enzyme, glucocerebroside builds up inside cells over time, making them get bigger. These enlarged cells are called Gaucher cells, and can lead to body organs to swelling, which causes the symptoms of Gaucher.

Simple science

Enzyme – things that help speed up chemical reactions.

Glucocerebroside builds up specifically in the recycling centres of ‘**rubbish collector**’ cells in the body.

The ‘rubbish collector’ cells are called macrophages and they have a hunger for waste! Macrophages are white blood cells that eat dead cells and germs to remove waste and fight infection.

All cells, including macrophages, have **recycling stations** to help break down, digest and recycle waste products produced by chemical reactions. These are called lysosomes and are like tiny stomachs within cells!

Who inherits Gaucher?

- Gaucher is caused by an altered gene that changes how much glucocerebrosidase enzyme cells make and how well it works breaking down glucocerebroside.
- To be diagnosed with Gaucher and to develop symptoms, a person must have two copies of the altered gene – one from each parent. The need to have two copies of a gene to develop a condition makes Gaucher a 'recessive' condition.
- People with one copy of the altered gene are known as 'carriers' and do not have Gaucher and won't experience any symptoms.

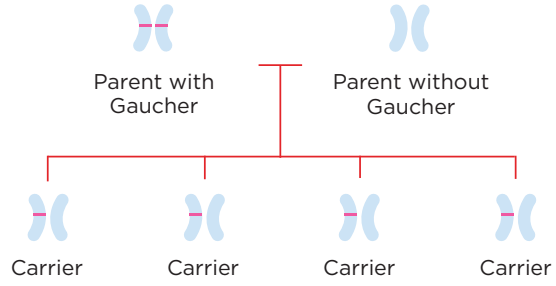
Decoding Gaucher

Alterations in the glucocerebrosidase beta 1 (*GBA1)* gene cause Gaucher. This gene tells cells how to make glucocerebrosidase in the recycling stations (lysosomes) of macrophage cells.

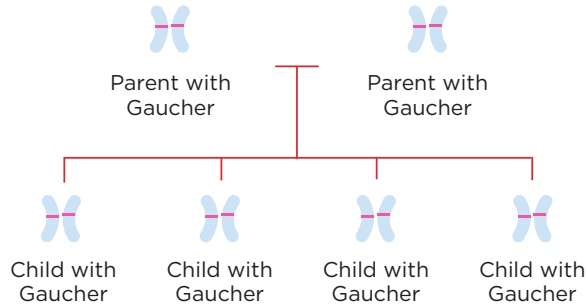
The *GBA1* gene can be altered in hundreds of different ways, meaning that different people can produce differing amounts of the enzyme.

Some produce a moderate amount, some very little, and some produce none at all. The lower the amount of glucocerebrosidase produced, the greater the build-up of glucocerebroside, which can cause damage to cells and organs in the body.

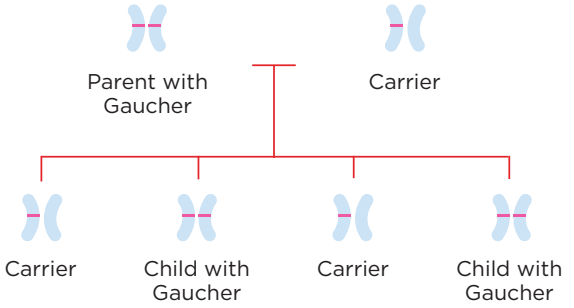
One parent with and one without Gaucher



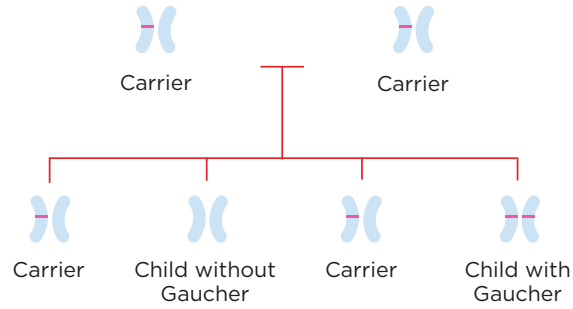
Both parents have Gaucher



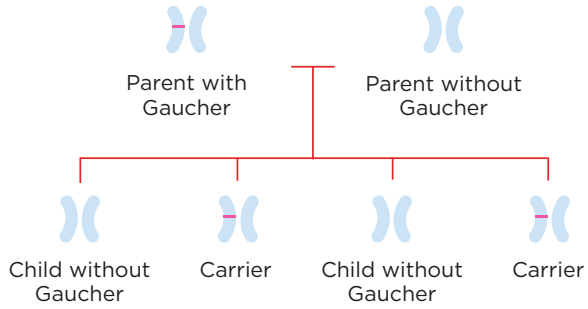
One parent with Gaucher and one is a carrier



Both parents are carriers



One parent is a carrier, the other is not



If both parents have Gaucher, all children will inherit Gaucher.

Full disclosure

This inheritance structure is used to demonstrate inheritance patterns only. Family structures can look very different.

Gaucher is rare!

Around 1 in 74,000 Australians have been diagnosed and are living with Gaucher

Simple science

There are three kinds of Gaucher:

Type 1 (non-neuronopathic)

- The most common form of Gaucher. 9 out of 10 people with Gaucher have this type.
- Symptoms vary and progress from childhood to adulthood.
- Typically affects the liver, spleen and bone marrow, and does not usually involve the nervous system.

Type 2 (acute neuronopathic)

- A severe form affecting the central nervous system in babies and infants.
- First symptoms occur around 3–6 months of age.

Type 3 (chronic neuronopathic)

- A chronic form affecting the central nervous system from childhood to adulthood.

This booklet covers the symptoms and experiences associated with **type 1 Gaucher**.

Gaucher symptoms

Gaucher is different for everyone. Symptoms can vary considerably from person-to-person and over time.

Some people have mild symptoms that start later in life; others have more severe symptoms that start in childhood.

No two people experience Gaucher in the same way.

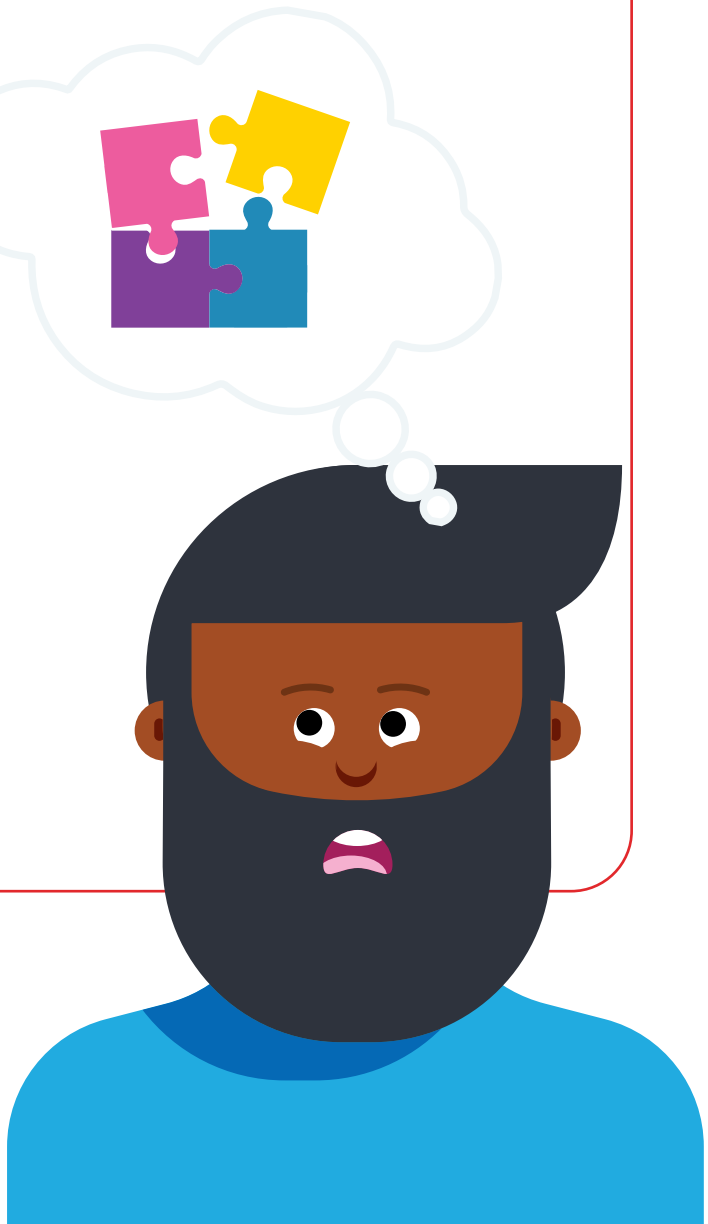
Fitting the pieces of the puzzle together

Gaucher is a difficult puzzle to crack!

Symptoms can be:

- **vague** and can overlap with those of other, more common conditions.
- **variable** from person to person. They can range from mild to severe and can affect different organs, making a diagnosis more complicated.

As a rare disease, Gaucher is not always top of mind for doctors, as they don't see it very often. This means it can take some time for people to be diagnosed.



Symptoms

Bleeding
Some people may have nose bleeds, gum bleeding, or bleeding after surgery or dental procedures.

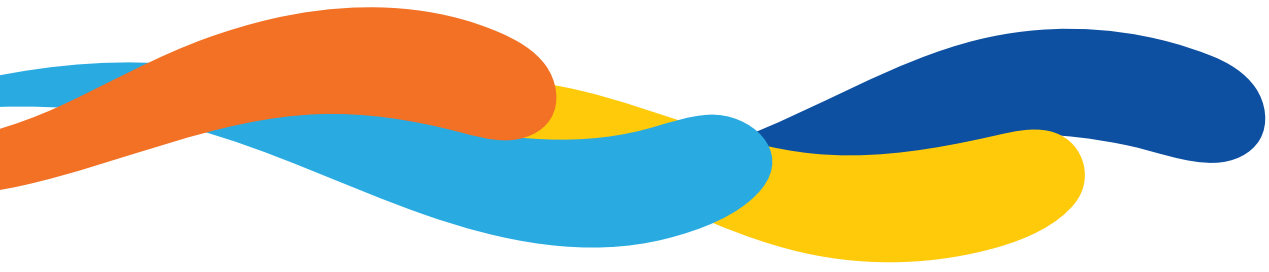
Low levels of certain blood cells
With the build-up of glucocerebroside in the cells, it may be difficult for the body to produce and maintain blood cell counts. People may have low levels of red blood cells (anaemia) and cells that help blood to be able to clot after an injury (thrombocytopenia).

Bruising
Occurs due to low levels of cells that help blood clot, known as platelets (thrombocytopenia).

Extreme tiredness (fatigue)
Some people may experience shortness of breath, weakness and tiredness due to low levels of red blood cells (anaemia).

Swollen belly (enlarged liver or spleen)
The build-up of glucocerebroside in cells may cause organs such as the spleen or liver to become larger than usual. This may put pressure on the stomach causing swelling and pain. People may also find it difficult to eat a full meal.

Bone pain and weakening
Bones may deteriorate and become thinner, making them painful or weak and sometimes easier to break.



Symptoms that may progress

Over time, people with Gaucher may develop:

Weak and fragile bones (osteoporosis)

Kidney symptoms

Heart symptoms

Lung symptoms

In some cases, Gaucher can cause serious complications for the bones, kidneys and heart. People with Gaucher or their care partners are advised to discuss any questions or concerns around symptoms with their healthcare team.

What causes Gaucher symptoms?

Gaucher symptoms are the result of the gradual build-up of the fatty substance glucocerebroside in the body's cells and tissues.

Without treatment, the build-up can result in damage, especially to the kidneys and heart.

The good news is, treatments are available.

Available treatments

Treatments are available for Gaucher, including options to help manage symptoms.

With treatment, people with Gaucher may be able to lead full lives and carry out their usual daily activities.

Treatments to manage Gaucher symptoms

Gaucher is a difficult puzzle to crack!

- Medicines and therapies are available to help manage pain, blood cell levels and bone mineral loss.
- Healthcare teams can provide further information and advice about available treatments most suitable for the symptoms experienced.

Treatments for Gaucher itself

Treatments specific for Gaucher are available.

Healthcare teams can discuss appropriate treatment options to suit each person's circumstances and preferences.

The future with Gaucher

Gaucher is a progressive condition and, while there is no cure now, treatment is available to help reduce the build-up of glucocerebroside that can cause damage to the body. Research continues to look for new treatments and ways to improve the quality of life for people living with Gaucher.

Working closely with a healthcare team helps identify suitable treatment options tailored to each person with Gaucher's specific needs and symptoms.

Information is also available for parents and guardians of people with Gaucher in the **Guide for parents** booklet on the beMi website at bemi.health

Find out more

For more information about Gaucher and its treatment options, individuals are advised to consult their doctor or another member of their healthcare team. You can also visit the following independent Australian organisations:

Gaucher Association of Australia and New Zealand
gaucheranz.com.au
info@gaucheranz.com.au

Rare Voices Australia
www.rarevoices.org.au
info@rarevoices.org.au

Rare Helpline
www.rareportal.org.au/rare-helpline

Additional booklets and animations can also be found on the beMi website at **bemi.health**

In the spirit of reconciliation, Takeda acknowledges the Traditional Custodians of Country throughout Australia, and their connections to land, sea and community. We pay our respects to their Elders past, present and emerging, and extend that respect to all First Nations peoples.

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