What is Fabry?

A guide for people living with Fabry disease

Takeda

beMi®

Inside this booklet

This booklet provides general information and is not a replacement for medical advice. Any questions about medical conditions, including Fabry, should be discussed with a healthcare professional.

What is Fabry?

- Fabry disease is a genetic condition that is passed down from parents.
- Fabry is caused by an alteration or change in a certain gene.
- As a result of this change, an important enzyme called alpha-gal A does not work in the way as it should.
- Alpha-gal A helps break down a type of fat (Gb3) in the recycling centres of cells throughout the body.
- Without enough alpha-gal A, Gb3 builds up over time, which can cause damage to cells and organs in the body.
- Fabry is a life-long, progressive condition. However, treatments are available.

Simple Science

Alpha-gal A's full name is alpha-galactosidase A.

Enzyme – things that help speed up chemical reactions.

Gb3's full name is globotriaosylceramide.

The recycling stations in our cells help break down, digest and recycle waste products produced by chemical reactions. These are called lysosomes and are like tiny stomachs within our cells!

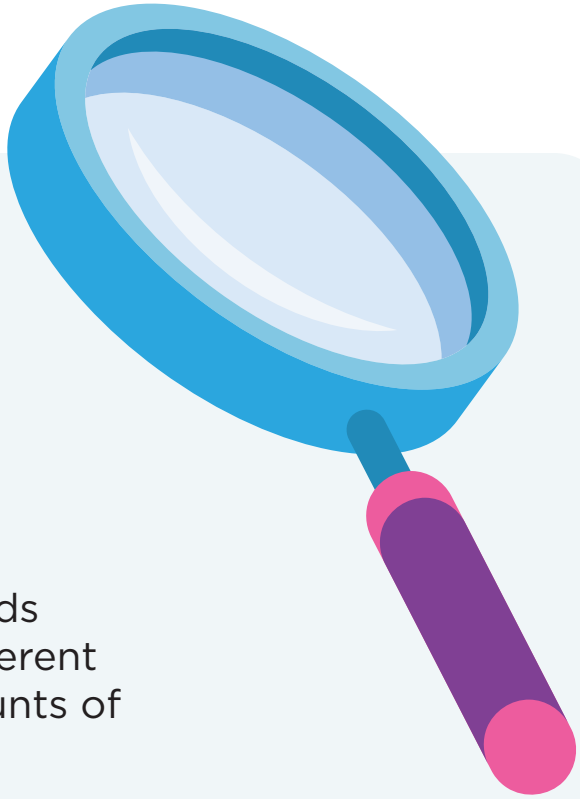
How do people get Fabry?

Fabry is caused by an altered gene that changes how much alpha-gal A cells make and how well it works breaking down Gb3.

This gene is found on the X chromosome – one of the two chromosomes we inherit from our biological parents.

We each inherit:

- an X chromosome from a female parent
- either an X or a Y chromosome from a male parent, which determines sex assigned at birth.

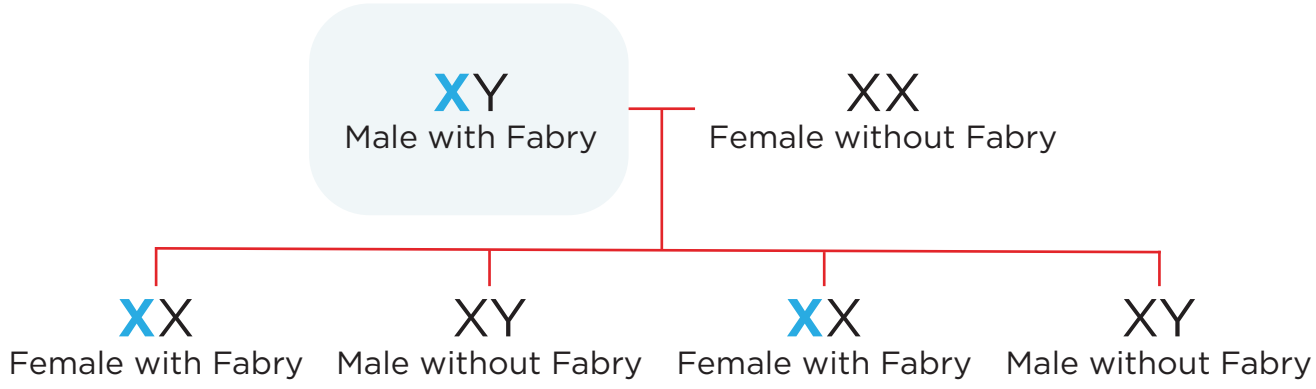


Decoding Fabry

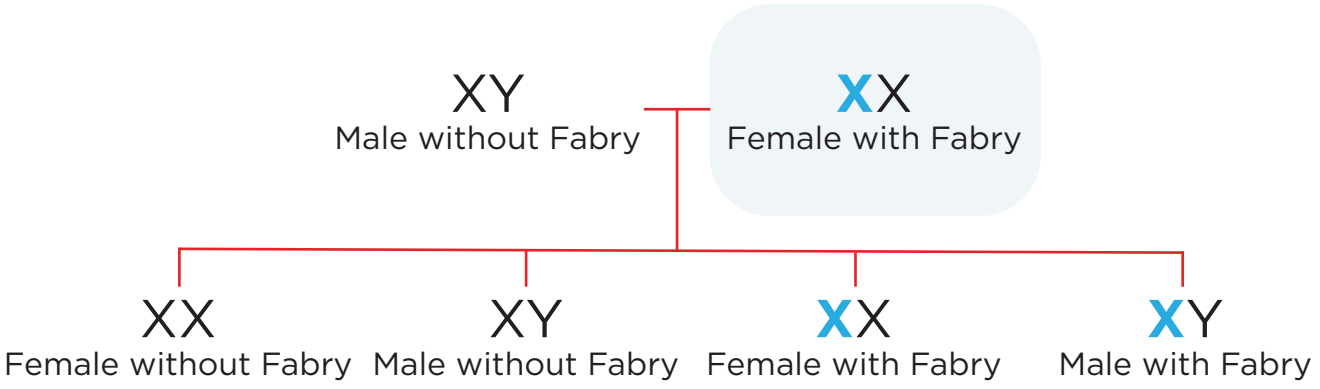
Alterations in the *GLA* (galactosidase alpha) gene cause Fabry. This gene tells cells how to make alpha-gal A in the recycling stations (lysosomes) within cells. This gene can be altered in hundreds of different ways meaning that different people can produce differing amounts of the alpha-gal A enzyme.

Some produce a moderate amount, some very little, and some produce none at all. The lower the amount of alpha-gal A produced, the greater the build-up of Gb3, which can cause damage to cells and organs in the body.

Male parent has Fabry



Female parent has Fabry

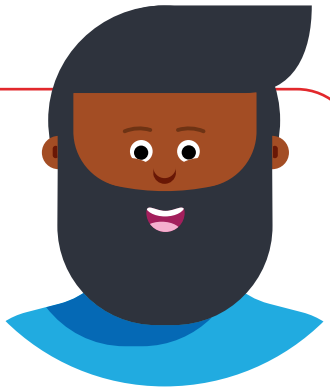


Full disclosure

This inheritance structure is used only to demonstrate inheritance patterns. Family structures may vary from this example.

The terms ‘male’ and ‘female’ refer to genes inherited and the sex assigned at birth. For simplicity, this refers only to X and Y chromosomes. However, the inherited genes can differ for individuals who are intersex.

Fabry is rare! Around 500 Australians have been diagnosed and are living with Fabry



Fabry in males

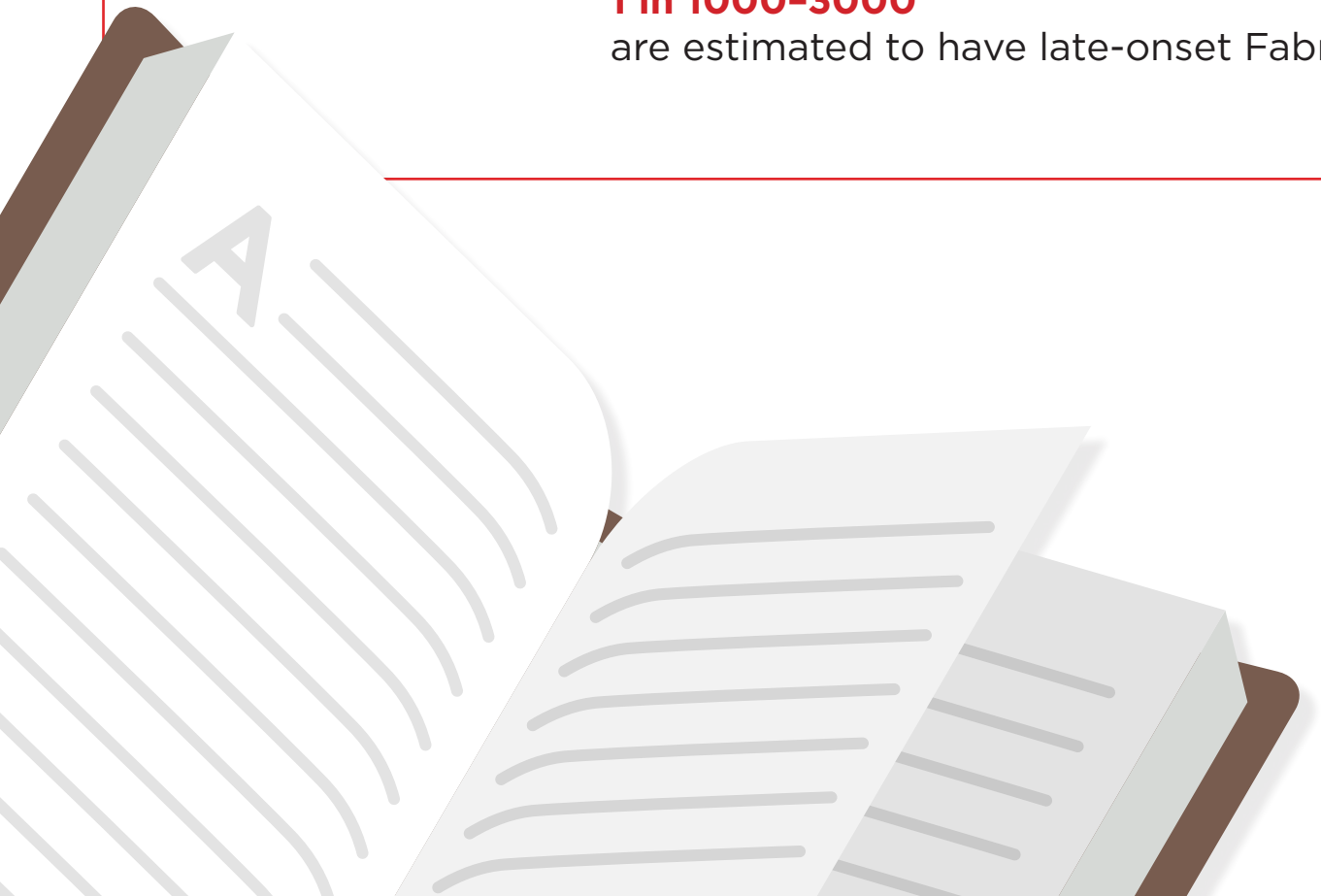
Sex chromosomes: XY

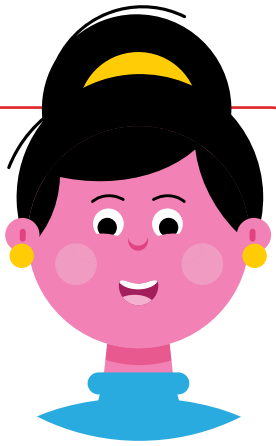
Males have only one X chromosome and will only develop Fabry if they inherit the X chromosome with the Fabry gene.

Symptoms tend to be more severe in males because the X chromosome with the Fabry gene operates in every cell.

1 in 22,000–40,000
males are estimated to have the classic form of Fabry.

1 in 1000–3000
are estimated to have late-onset Fabry.





Fabry in females

Sex chromosomes: XX

Generally, only one of the two X chromosomes will have a copy of the Fabry gene.

Symptoms vary from mild through to severe depending on whether the Fabry or non-Fabry X chromosome is operating in cells – it can be different from cell to cell.

1 in 6000–40,000
are estimated to have later-onset Fabry.

There are two kinds of Fabry:

Classic

- low levers or very little activity of alpha-gal A
- severe with symptoms developing in childhood or adolescence

Later onset

- some alpha-gal A activity
- symptoms begin after 30 years of age
- symptoms usually milder and progression may be slower than the classic form

Simple Science

Fabry symptoms

Symptoms can vary over time, or even from day to day.

Some people have mild symptoms that start later in life, while others have severe symptoms that begin in childhood.

Everyone experiences Fabry disease differently.

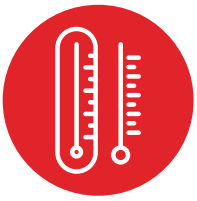
Fitting the pieces of the puzzle together

Fabry is a difficult puzzle to crack!

- Symptoms of Fabry can occur anywhere in the body because of the build-up of Gb3 in nerve cells, blood vessels and organs.
- Symptoms can be vague and common to many different conditions seen more frequently than Fabry.
- Symptoms are different in different people, unlike other health conditions.
- It is a rare condition, so doctors don't see it often.

As a result, it can take some time for people to be diagnosed.

Early symptoms



Reduced ability to sweat
Can cause a person to feel hot or have difficulty tolerating heat.

Pain or discomfort in hands and feet
This may occur during exercise or in hot weather.

A swirl-like pattern on the front surface of the eye
An optometrist may notice this during an eye test. About three out of four people with Fabry disease have this sign.

A spotty, dark red rash
Often appears between the belly button and knees.

Gut symptoms
Such as abdominal pain, diarrhoea, constipation, nausea or vomiting.

Later symptoms

Feeling tired or fatigued

Headaches or feeling dizzy

Hearing loss or ringing in the ears

Chest pain or a racing heartbeat

Kidney problems

In some cases, Fabry can cause serious complications for organs such as the kidneys, heart and brain.

Consult a doctor if there are concerns about any symptoms.

What causes Fabry symptoms?

Fabry symptoms are the result of the gradual build-up of the fatty substance Gb3 in the body's cells and tissues.

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

The good news is, treatments are available.

The future with Fabry

Fabry is a progressive condition and, while there is no cure now, treatment is available to help reduce the build-up of Gb3 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 Fabry.

With a Fabry diagnosis, individuals can work with their healthcare team to find the best treatment for their needs and symptoms. For more information, see the ***Living with Fabry*** booklet on the beMi website.

What treatments are there for Fabry?

Treatments for Fabry can be divided into those that help manage symptoms, and those that treat the condition itself.

Treatments to manage Fabry symptoms

- For example, medicines to help manage pain and burning sensations, stomach problems, high blood pressure (hypertension), kidney problems and high cholesterol.
- Hearing loss can be treated with hearing aids.

Treatments for Fabry itself

Treatments specific to Fabry are available. Consult a doctor to discuss which treatment is best.

Find out more

To find out more about Fabry or its treatment, people should consult their doctor or another member of their healthcare team. They can also visit the following independent Australian organisations:

Fabry Australia
www.fabry.com.au
admin@fabry.com.au

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

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

You can also find other booklets and animations 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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