This leaflet is for people who have been diagnosed with Fabry disease

Fabry disease & your family

Find out how Fabry runs in families and what you can do to support your relatives

Disclaimer: This brochure has been funded by Amicus Therapeutics.
It is designed to help you talk to family members about Fabry disease.
It does not in any way suggest a diagnosis of Fabry disease for any individual.
Any concerned individual should speak to a healthcare professional.

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Your **Fabry diagnosis** can help your family

As you may have experienced, the journey to a Fabry diagnosis can be long and difficult, often involving many doctors, appointments and tests.[1,2]

Now that you have received a diagnosis of Fabry, you may be able to help other family members find out if they have Fabry too.

How can your diagnosis help your relatives?

You may already know that Fabry is a genetic disorder which can be passed down the generations of a family. But did you know that on average, for each person with Fabry, at least five other members of their family can be diagnosed too?[3]

Now that you know about your Fabry, this means that you could have children, parents, siblings, aunts, uncles or cousins that also have Fabry but don't yet know. You could help them.

You may have family members with Fabry who are not experiencing symptoms yet.[4] Others may already be experiencing symptoms but may not realise what the cause is. It is important to know that their experience of symptoms may be different to yours.[4-7]

Each person with Fabry can have different symptoms, and these are often similar to other more common conditions. This means it can take a long time to be given the right diagnosis.[1,2,8] Being tested for Fabry or even knowing that Fabry is in the family could help someone get their diagnosis sooner.

This is important because Fabry is progressive, meaning it may get worse over time. With a diagnosis, they can get the support they need to help manage their condition, which may include treatment.[1,9-11]

Knowledge is power

By making sure your family are aware of your Fabry diagnosis, and letting them know that Fabry runs in families, you could potentially help other relatives get support and treatment.

Telling your family about your Fabry diagnosis may be daunting, so we have made this booklet to help. If you or your family have any questions about Fabry, speak with your healthcare professional.

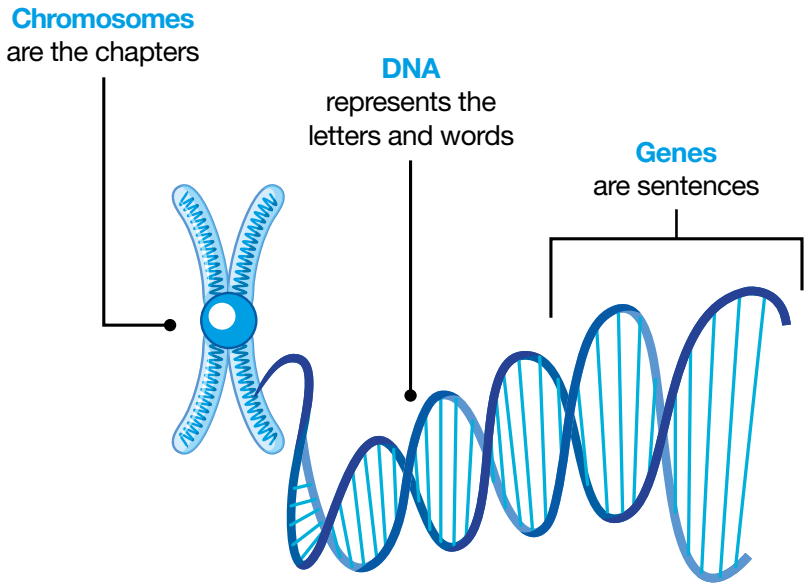
If you have any questions about Fabry disease, speak to your healthcare team.

What causes **Fabry?**

A beginner's guide to genetics[12,13]

Every cell of your body is programmed to work in a particular way, for example to help with digestion, keep your heart beating, or fight infections. To do its job properly, every single cell needs a set of instructions. DNA is 'read' by the cell and provides these instructions. A gene is a section of DNA that programmes one specific instruction. The genes themselves are grouped into separate units called chromosomes.

You can think of this like an instruction manual where:



Sometimes there are changes in the DNA. You can think of these like misspelt words – one wrong letter can completely change the meaning of the word. This can mean that the gene cannot be read by the cell, so the cell can't do its job properly.

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The genetics of Fabry[1,4]

Fabry is caused by changes in a gene called GLA which provides the instructions to make an enzyme (α-galactosidase A). Normally this enzyme helps break down certain sugary-fatty substances (glycosphingolipids) in the cells of our body – but the changes in the gene mean it can't do its job. This leads to a build-up of those substances causing the symptoms of Fabry.

Having a change on the Fabry gene means that the body's cells can't function properly, and that may lead to the symptoms of Fabry.

If you have any questions about Fabry disease, speak to your healthcare team.

How **Fabry** runs in families

The X and Y chromosomes determine the sex of a person:[14]

Females have two X chromosomes
– sons and daughters will randomly receive one of these X chromosomes

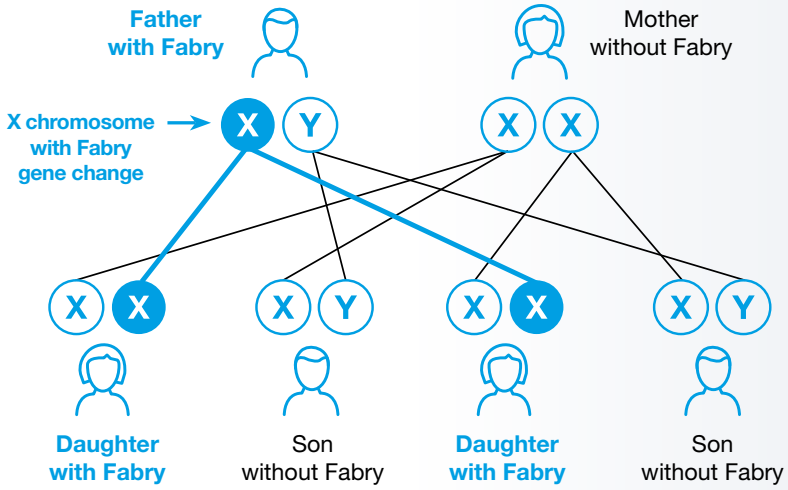
Males have one X and one Y chromosome
– sons will receive the Y chromosome and daughters the X chromosome

The GLA gene, which is changed in Fabry, is located on the X chromosome and therefore Fabry is referred to as an "X-linked disorder".[14]

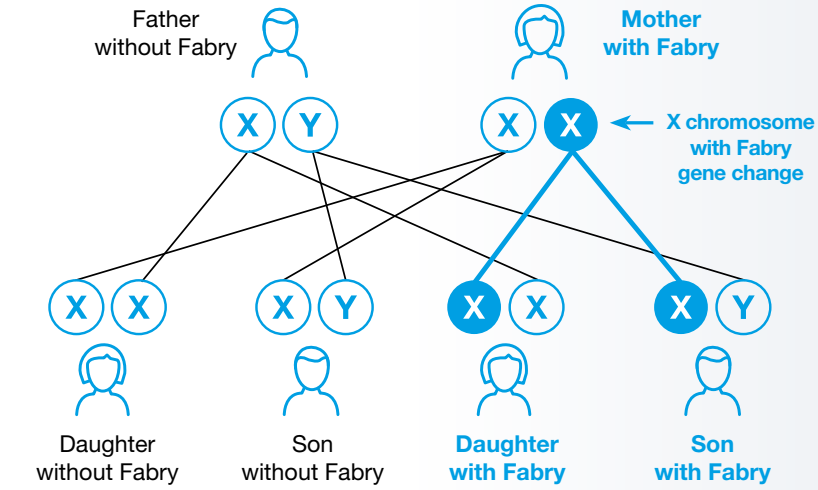
Males and females can both have Fabry but the likelihood of a father or a mother passing down the Fabry gene change is not the same.[7,14] It depends on the chromosomes they pass onto their children.[14]

If you have any questions about Fabry disease, speak to your healthcare team.

A father with Fabry will pass the Fabry gene change onto all of his daughters but none of his sons[14]



A mother with Fabry has a 50:50 chance of passing the Fabry gene change onto any of her daughters or sons[14]

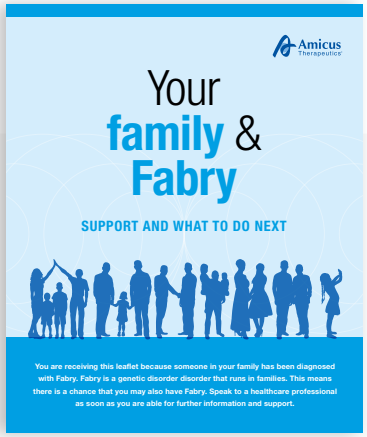


Starting conversations with your family

It is important to understand that other people in your family may have Fabry without knowing it. So, the next step is for you to have a conversation with your relatives to explain that you have Fabry and they can talk to their healthcare professional if they want to find out more.

If you are unsure, a member of your healthcare team can advise you on how to approach the situation.

You can give your relatives a copy of the **'Your family and Fabry'** leaflet so they can read about Fabry and decide that they would like to do next.



Speaking to your family members about Fabry means they can decide what they want to do next.

What should your family do next?

If your family members would like to find out more about Fabry, the next step for them is to go and see a healthcare professional. They might then be referred to a specialist for further testing.

- They can then have a discussion with a specialist who can assess their potential risk of having Fabry and discuss options for testing.
- If they decide to be tested for Fabry, these tests are usually performed on a cheek swab, blood or other tissue sample.[15] Tests will be performed to look for changes to the GLA gene which may cause Fabry disease.[4]
- If a Fabry gene change is found, their medical team will discuss more about the disease, how to manage it and any potential implications.
- Treatment options are available for Fabry disease, and your specialist will discuss whether these are suitable.

If you have any questions about Fabry disease, speak to your healthcare team.

Key points

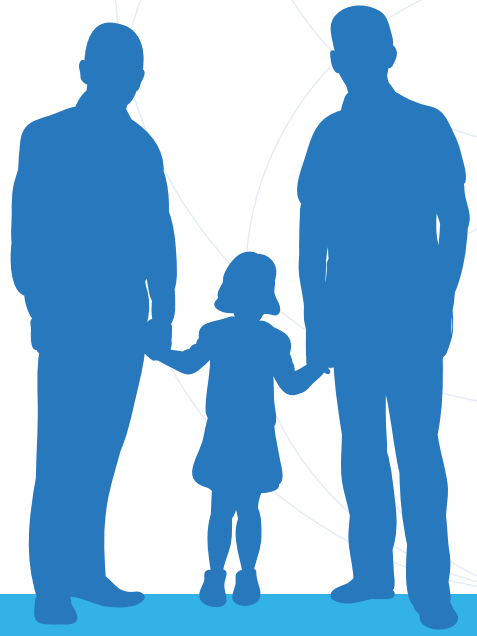
Here's a summary of the important messages from this booklet:

1. Fabry is a genetic disorder which runs in families.
2. Having a change on the GLA gene means that the body's cells can't function properly, and that may lead to the symptoms of Fabry.
3. Now that you have received a diagnosis of Fabry, you may be able to help other family members find out if they have Fabry too.
4. Some family members may have Fabry without knowing it.
5. They can speak to a healthcare professional to find out more about Fabry and options for testing.

Helpful resources

If you have any questions about Fabry, you can speak to your healthcare team.

[For Affiliates - Recommend including URL for Amicus Fabry Family Screening website if available /localised]



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