# A visual guide to understanding Fabry disease

**Information for people living with Fabry disease - and their families**

Plain English Campaign's Crystal Mark for clarity does not apply to the design of this guide.

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Crystal Mark 23941
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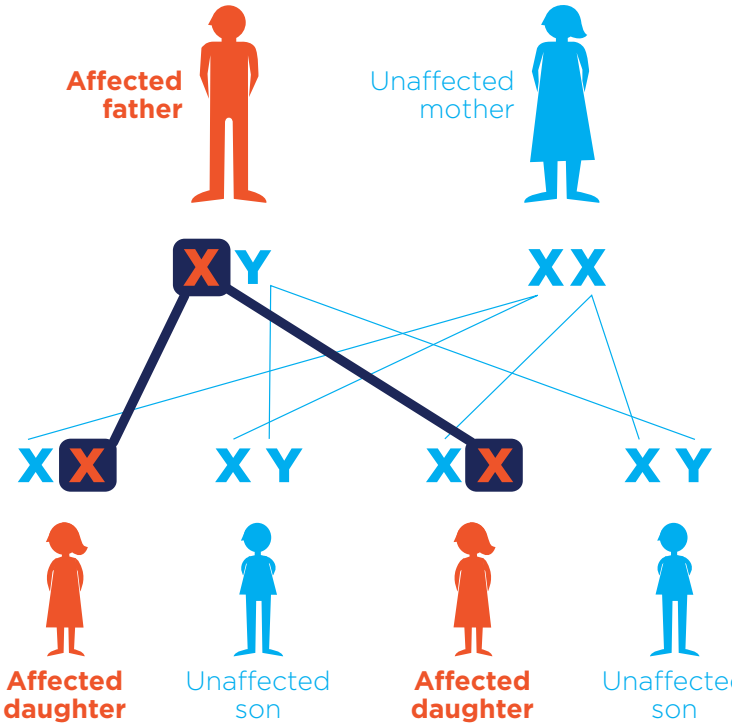
## How is Fabry disease passed on?

- Males have an X chromosome (inherited from their mother) and a Y chromosome (inherited from their father).
- Females have two X chromosomes (one inherited from their mother and one from their father).
- Fabry disease is an X-linked disorder, meaning that it is passed on in a mutation in an X chromosome. It can be passed down by either parent if they have a mutated X chromosome.

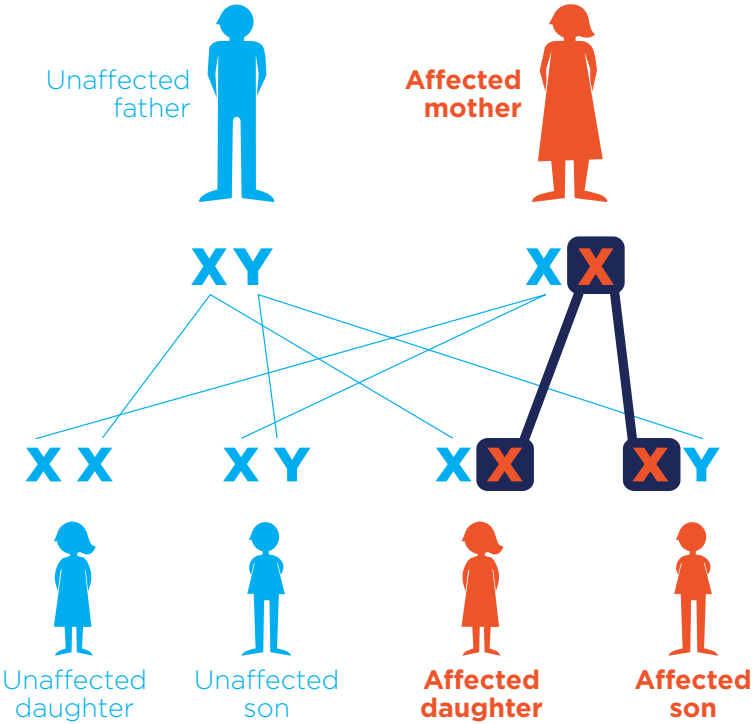
In the following diagrams, mutated X chromosomes are shown in red.
Any child who inherits a mutated X chromosome will develop Fabry disease.

### How fathers pass on Fabry disease




As shown in the diagram above, if a father has the mutated X chromosome, any daughter will develop Fabry disease but sons will not.

### How mothers pass on Fabry disease




As shown in the diagram above, if a mother has the mutated X chromosome, there is a 50% chance of any child (male or female) developing Fabry disease.

## What do certain words mean?

### A glossary of important terms when discussing Fabry disease

| *Cell* | *Chromosome* | *DNA (deoxyribonucleic acid)* | *Enzyme* | *Lysosomal disorder (LD)* | *Mutation* |
|---|---|---|---|---|---|
| A basic building block of all living things. | A structure that contains DNA and a person's genetic code. | A basic unit that carries genetic information from one generation to the next and contains instructions (referred to as code) for making proteins and enzymes. | A special type of protein that speeds up a reaction that takes place in the body. | A disorder of lysosomes (specialised fluid-filled sacs found in cells) resulting in an accumulation of waste products in the cell. | A permanent error in the DNA code. |

## Facts about Fabry

(For more information, talk to your healthcare provider.)

Fabry is a rare, progressive, genetic disease. It is thought to affect 1 in 117,000 people, but it may be more common than this.

It's a type of disease called a lysosomal disorder.

People with lysosomal disorders have problems making specific enzymes.

In Fabry disease, the affected enzyme is 'α-galactosidase A', referred to as α-Gal A.

Usually, α-Gal A breaks down substances called globotriaosylceramide (GL-3) and globotriaosylsphingosine (lyso-Gb3) that are in cells.

In people with Fabry disease, α-Gal A does not break down GL-3. So GL-3 builds up, particularly in cells lining blood vessels.

This build-up damages tissues and organs, leading to the symptoms of Fabry disease.

## What causes Fabry disease?

Everyone has DNA, which is inherited from their parents.

Sometimes, there are mutations in the DNA code that makes up a particular gene.

Think of DNA code like spelling. One wrong letter can completely change the meaning of a word!

CAR

CAT

Some people with the mutation that causes Fabry disease make very little or no α-Gal A.

Some people with the mutation that causes Fabry disease may make α-Gal A that doesn't function correctly.

Every patient is **unique**

## How Fabry disease could affect you

**Everyone experiences Fabry disease differently, but the following are common.**

**Eyes**
- A pattern of hazy rays in the cornea
- Cataracts

**Heart**
- Irregular heartbeat (fast or slow)
- Heart attack or heart failure
- Enlarged heart

**Kidneys**
- Protein in the urine
- Decreased kidney function
- Kidney failure

**Skin**
- Sweating less than or more than normal
- Small dark red spots called angiokeratomas, particularly over the buttocks, groin and lower abdomen

**Nervous system**
- Pain
- Hearing loss, ringing in the ears
- Intolerance to heat, cold or exercise
- Transient ischaemic attack (mini stroke) and stroke
- Feeling dizzy

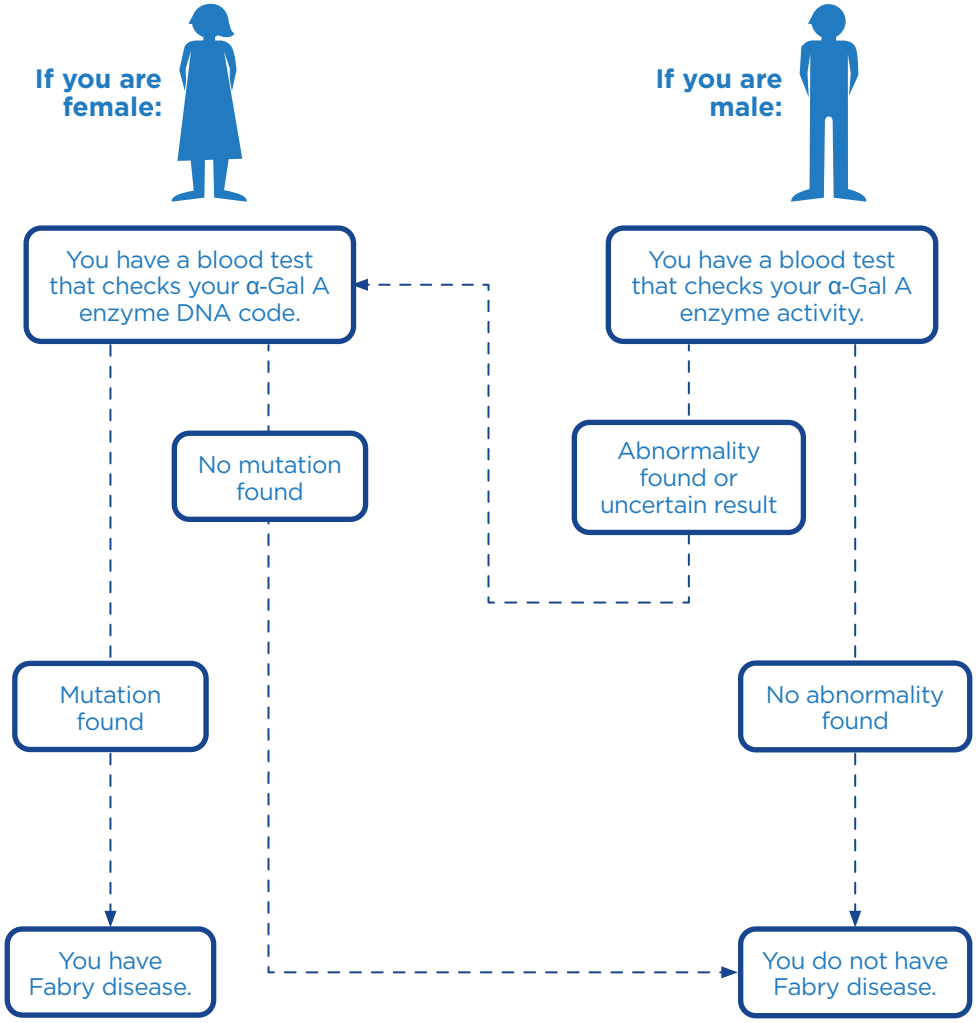
**Gastrointestinal**
- Nausea, vomiting, cramping and diarrhoea
- Pain or bloating after eating, feeling full after a small amount of food
- Constipation
- Difficulty managing weight

**Being diagnosed with Fabry disease may have psychological effects. These include:**
- guilt, apprehension, depression, isolation and even fear
- relief at finally getting a diagnosis of Fabry, after years of not knowing, and shock

**Bottom line:**

Talk to others with Fabry disease and share your experiences. It may help!

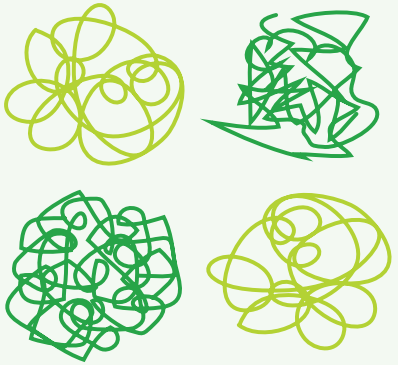
## If a doctor suspects Fabry disease, here's what might happen




- Your doctor may suggest **genetic counselling**.
- Your doctor begins to create a **personalised approach** to your monitoring and treatment.
- Your **family history** is taken.
- Your doctor may suggest genetic testing for **other family members** at risk of having the mutated chromosome.

## Why do mutations matter?


More than 1000 mutations have been identified in the gene linked to Fabry disease.



The type of mutation may help predict **when** symptoms appear, **what kind** of symptoms appear, and **how bad** the symptoms are or may become.

It's important to know **which mutation** you have.

**If you want to learn about Fabry and its effects, contact your local patient organisation or talk to your healthcare professional.**

**Information in this guide came from the following sources.**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