



X-linked recessive inheritance: carrier mother

This communication aid has been produced for clinicians to help support and guide conversations about X-linked recessive inheritance with their patients.

What causes X-linked recessive conditions?

We all have over 20,000 genes, which provide instructions for how our body works.

Our genes are packaged into structures called **chromosomes**. Our chromosomes come in pairs, with one inherited from each parent.

Most chromosomes (the autosomes) are numbered 1–22. Our sex chromosomes are called X or Y, and usually determine sex assigned at birth.

- Females typically have two X chromosomes.
- Males typically have one X and one Y chromosome.

X-linked recessive conditions are caused by **changes in genes** that are located on the X chromosome.

XY



Males who have a change on their X chromosome would be expected to develop the condition.

XX



Females who have a change on one of their two copies of the X chromosome are known as carriers. Usually, they would not develop the condition. In some cases, they may develop a milder version of the condition.

How are X-linked recessive conditions inherited?

When a female carrier has a child with someone who is not affected by the condition, either the X chromosome with the change or the X chromosome without the change will be inherited by the child.

This means, for every pregnancy:

- There is a 25% (1 in 4) chance of having a female carrier child (like their mother).
- There is a 25% (1 in 4) chance of having a female unaffected child, who is not a carrier.
- There is a 25% (1 in 4) chance of having a male child affected with the condition.
- There is a 25% (1 in 4) chance of having a male child who is not affected with the condition.

Key terms

Chromosomes: Packages of DNA which are found in our cells.

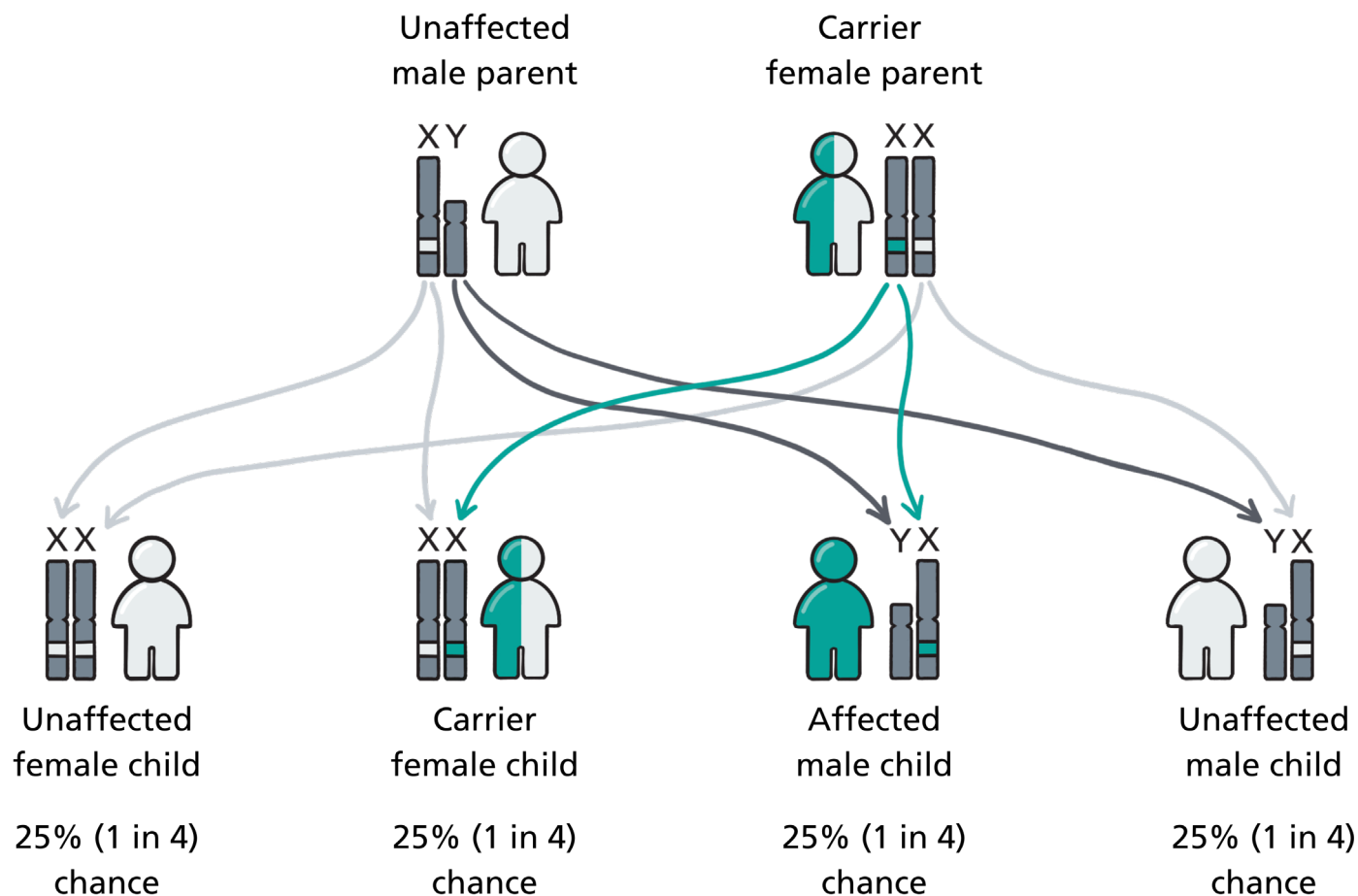
Gene change: Changes in a gene or chromosome used to be referred to as 'mutations'. Now, they are more commonly called changes, alterations or variants.





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Key

- X chromosome with a change
- X chromosome without a change
- Y chromosome

Want to learn more?

Scan to watch an animation explaining X-linked recessive inheritance



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Last reviewed: 14 February 2025