## WHAT CAN BE DONE IF YOU HAVE A X-LINKED CONDITION?

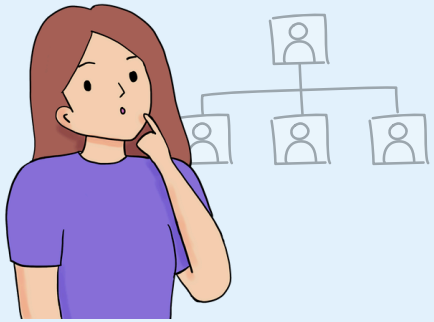
While many genetic conditions currently do not have a cure, genetic counselling is recommended, especially when planning for pregnancy. Genetic testing can provide valuable information about the risks and available options.

Depending on the genetic test results, the couples may consider:

- Prenatal diagnosis by fetal genetic analysis through chorionic villus sampling or amniocentesis when the couples conceive again.
- In vitro fertilization with special genetic testing called preimplantation genetic diagnosis (PGD) to select unaffected embryos.

## SHOULD I TELL YOUR FAMILY MEMBERS IF I HAVE AN INHERITED CONDITION?

If you have an inherited condition, it is important to inform your family members, as they may also have inherited the condition and could benefit from knowing about it. This is particularly crucial if they are planning to have children. It is recommended for them to seek advice from healthcare professionals to understand the implications and potential risks associated with the condition.



For further enquires, you may contact us.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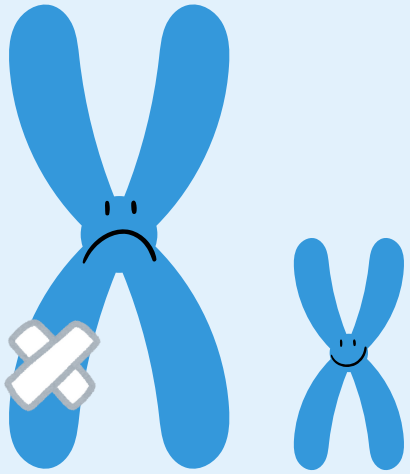
GENETICS AND GENOMICS CLINIC
DEPARTMENT OF CLINICAL GENETICS



# X-linked Inheritance

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# X-linked Inheritance

## WHAT ARE GENES AND CHROMOSOMES?

Genes act as instructions that guide the growth and functioning of the human body. They are located on structures known as chromosomes. In each cell, there are typically 46 chromosomes arranged in 23 pairs. The first 22 pairs, called autosomes, carry numerous genes responsible for determining human physical characteristics.

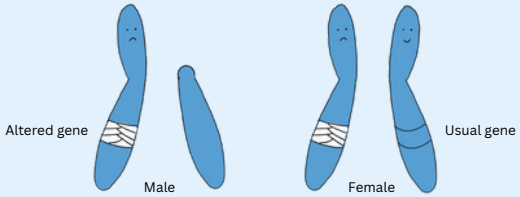
The last pair is sex chromosomes, which determine the sex of a human being. Females have two X chromosomes, with one inherited from each parent. On the other hand, males have one X chromosome inherited from the mother and one Y chromosome inherited from the father.

## WHAT DOES X-LINKED INHERITANCE MEAN?

X-linked inheritance is the way certain genetic conditions are passed down through the gene located on the X chromosome.

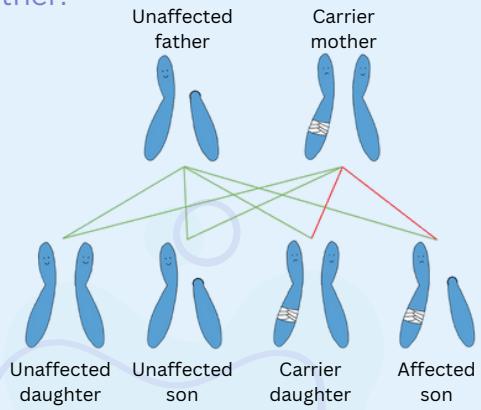
In X-linked inheritance, a female with an altered gene on one of the X chromosomes may remain a healthy carrier, because she has a second normal copy of the gene on her other X chromosome. However, there are some X-linked conditions where females can develop mild symptoms.

Males with an altered gene on the X chromosome would be affected because they have only one X chromosome, and therefore they would demonstrate traits of the condition.



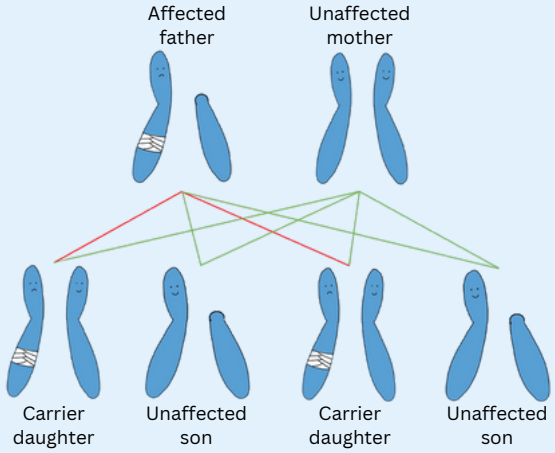

## HAVING CHILDREN (FEMALES)

If a woman is a carrier of an altered gene, there is a 50% chance that her sons will be affected by the condition, while her daughters would have a 50% chance of being a carrier like their mother.




## HAVING CHILDREN (MALES)

When men with X-linked conditions have daughters, all of them will be carriers of the altered gene. As men do not pass on their X chromosome to their sons, all the sons will be unaffected and will not be carriers.




## WHAT IS X-INACTIVATION?

X-inactivation is a natural process in which females randomly "turn off" one of their X chromosomes in each cell to balance gene expression. This process occurs naturally to stop too many X chromosome genes from being active. However, in some cases, X-inactivation can be skewed or unbalanced, leading to female carriers with affected conditions who have a higher proportion of cells expressing the altered X chromosome, resulting in clinical symptoms.