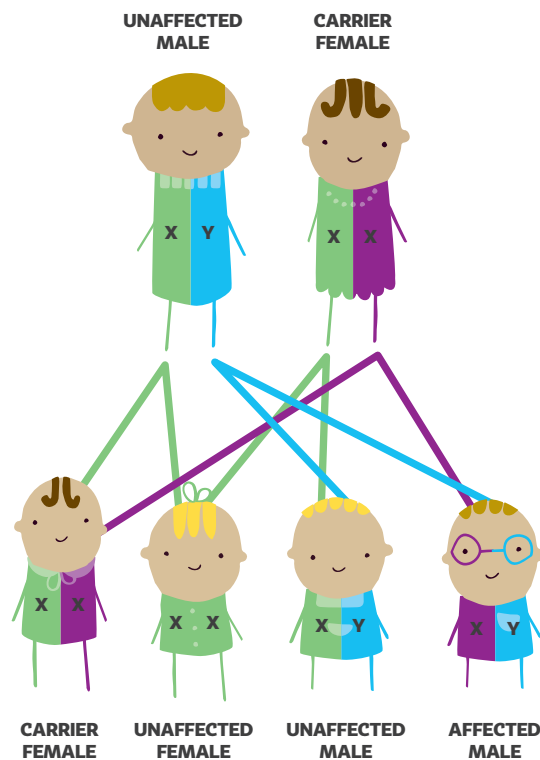


# WHAT IS X-LINKED INHERITANCE?

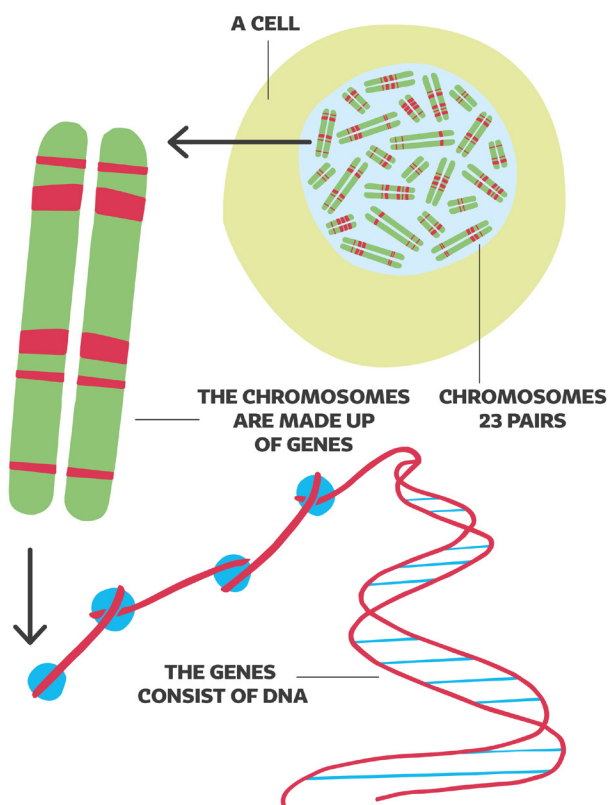
This leaflet will give you information about what X-linked inheritance means and how X-linked conditions are inherited.

## HOW X LINKED RECESSIVE CONDITIONS ARE PASSED ON BY FEMALE CARRIERS



## Genes and chromosomes

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are also responsible for many of our characteristics, such as our eye colour, blood type and height. We have thousands of genes. We each inherit two copies of most genes, one copy from our mother and one copy from our father. This is why we often have similar characteristics to both of them.

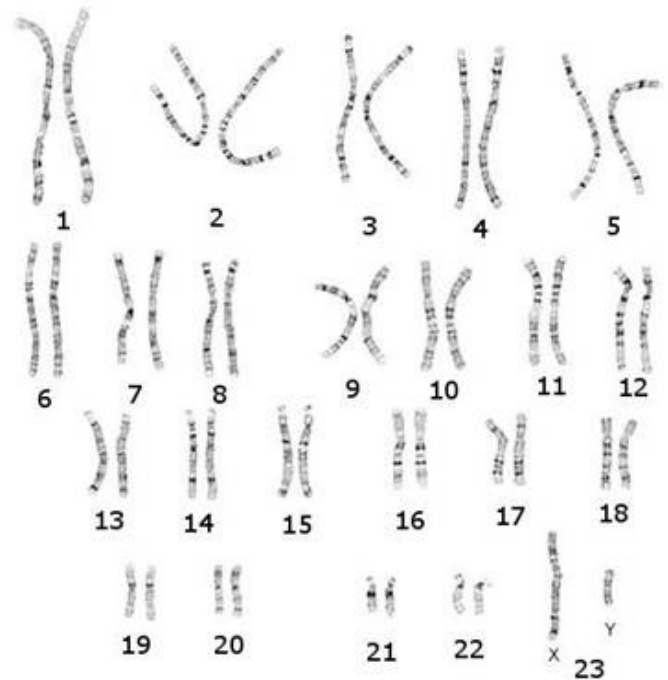


This picture shows a cell, DNA and chromosomes.

**Genes are located on small thread-like structures called chromosomes.**

Usually we have 46 chromosomes in most cells, one set of 23 chromosomes

inherited from our mother and one set of 23 chromosomes inherited from our father. So we have two sets of 23 chromosomes, or 23 pairs.



This is a picture of chromosomes.

The chromosomes numbered 1 to 22 above look the same in males and females. These are called the autosomes. Pair number 23 is different in males and females and they are called the sex chromosomes.

There are two kinds of sex chromosome, one called the X chromosome and one called the Y chromosome. Females normally have two X chromosomes (XX). A female inherits one X chromosome from her mother and one X chromosome from her father. Males normally have an X and a Y chromosome (XY). A male inherits an X chromosome from his mother and a Y chromosome from his father. The picture above therefore shows the chromosomes of a male as the last pair of chromosomes (XY).

## **Sometimes, there is a change in one copy of a gene which stops it from working properly.**

This change can cause a genetic condition because the gene is not communicating the correct instructions.

## **An X-linked genetic condition is caused by a change in a gene on the X chromosome.**

### **What is X-linked recessive inheritance?**

The X chromosome has many genes that are important for growth and development. The Y chromosome is much smaller and has fewer genes.

Females have two X chromosomes (XX) and therefore if one of the genes on an X chromosome has a change, the normal gene on the other X chromosome can usually compensate for the changed copy. If this happens the female is a healthy carrier of the X-linked condition. Being a carrier means that you do not have the condition, but carry a changed copy of the gene. In some cases, females show mild signs of the condition.

Males have an X and a Y chromosome (XY) and therefore if one of the genes on the male's X chromosome has a change, he does not have another copy of that gene to compensate for the changed copy. This means that he will be affected by the condition.

Conditions that are inherited in this way are called X-linked recessive conditions. Some

examples of X-linked conditions include haemophilia A and Duchenne muscular dystrophy.

### **X-linked dominant inheritance**

Though most X-linked conditions are recessive, very rarely X-linked conditions can be passed on in a dominant way. This means that even though a female inherits one normal copy and one changed copy of the gene, the changed gene will be enough to cause the condition. If a male inherits a changed X chromosome then this would be enough to cause the condition because males only have one X chromosome. An affected female has a 50% (1 in 2) chance of having affected children (sons and daughters). All of the daughters of an affected male will be affected but all sons will be unaffected.

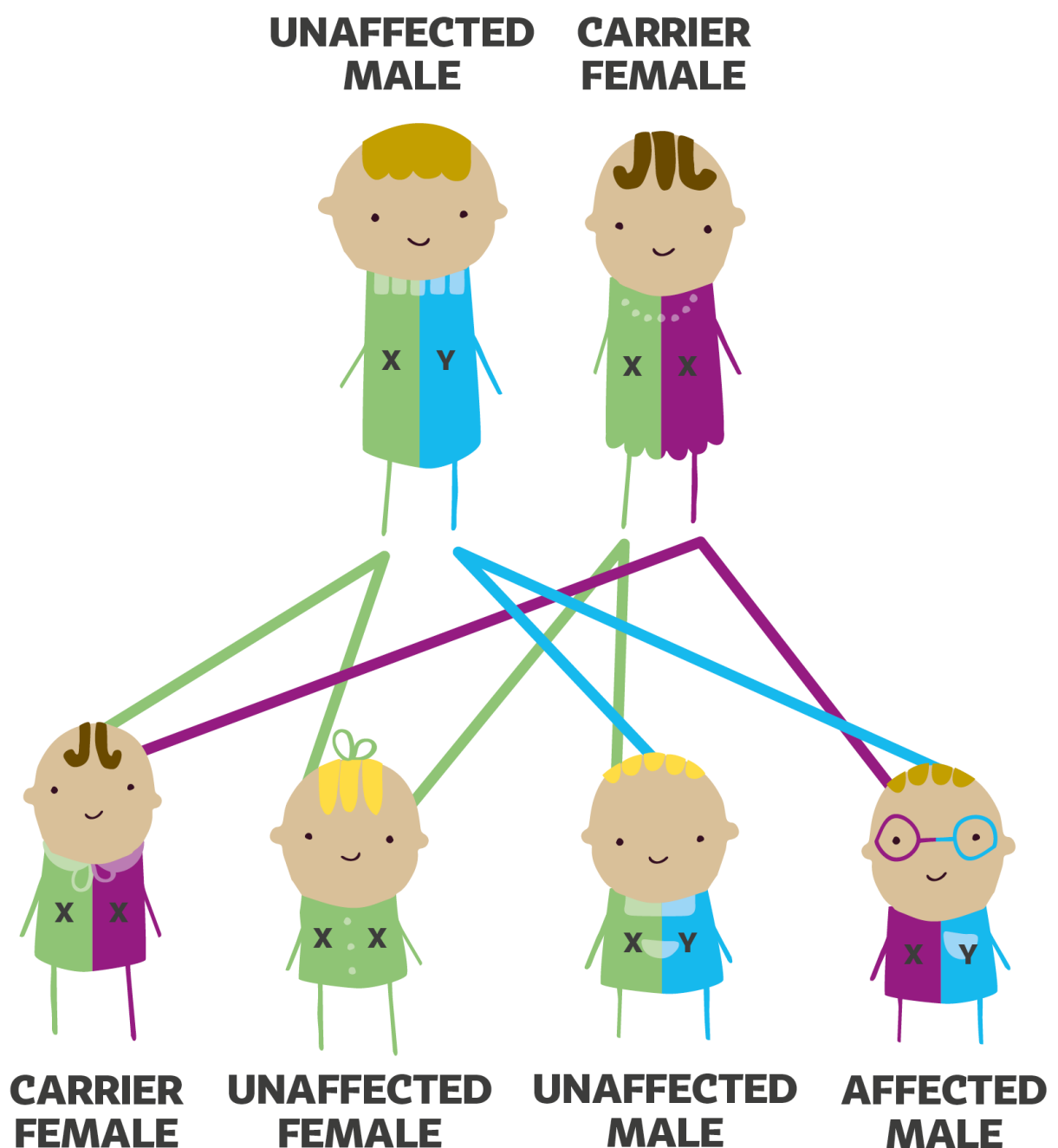
### **How are X-linked recessive conditions inherited?**

If a female carrier has a son, she will pass on either the X chromosome with the normal gene, or the X chromosome with the changed gene. Each son therefore has a 50% (1 in 2) chance of inheriting the changed gene and being affected by the condition. There is also a 50% (1 in 2) chance that the son will inherit the normal gene. If this happens he will not be affected by the condition. This chance remains the same for every son.

If a female carrier has a daughter, she will pass on either the X chromosome with the normal gene, or the X chromosome with the changed gene. Each daughter therefore has a 50% (1 in 2) chance of inheriting the changed gene. If this happens the daughter

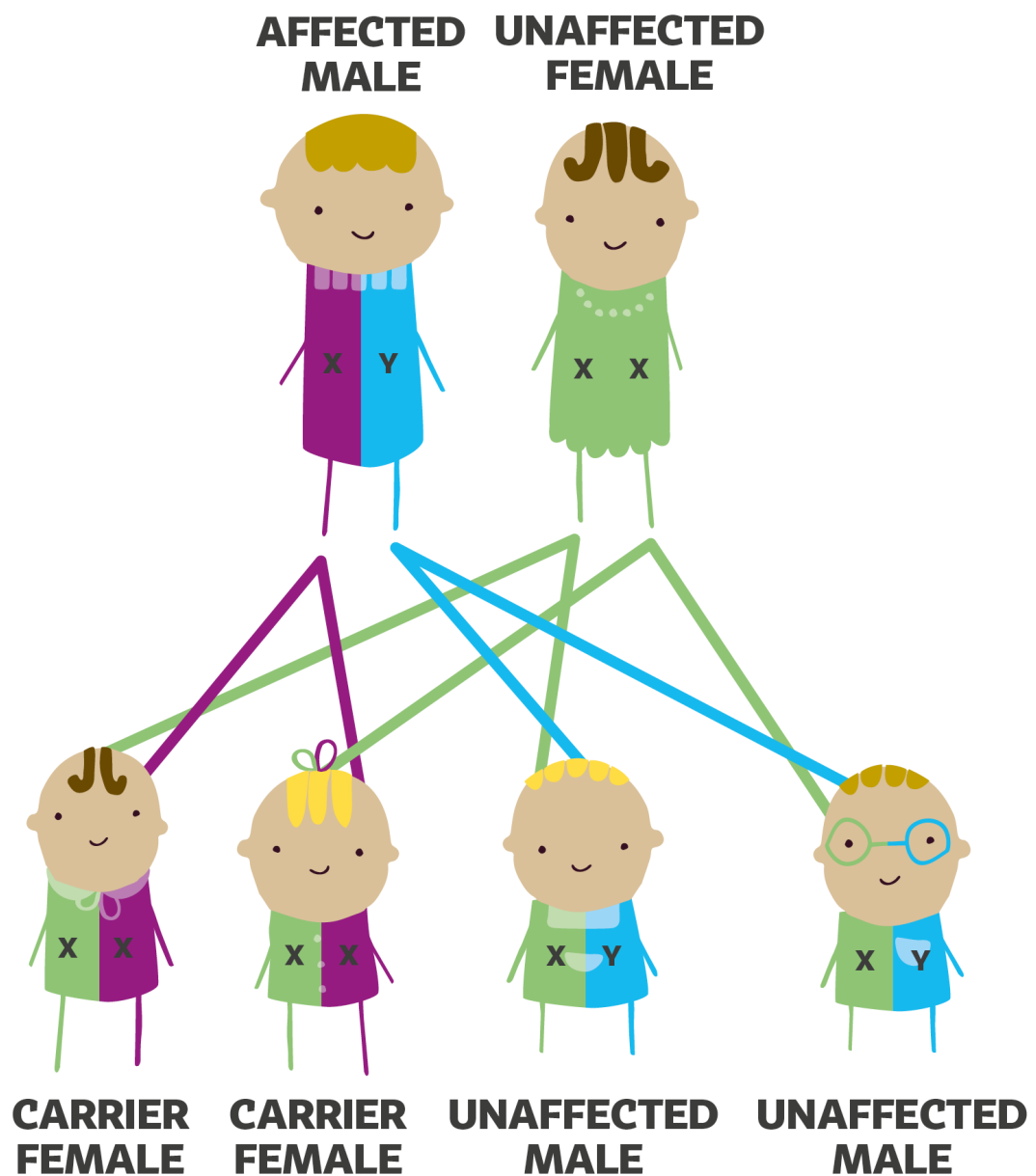
## HOW X-LINKED RECESSIVE CONDITIONS ARE PASSED ON BY FEMALE CARRIERS

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## HOW X-LINKED RECESSIVE CONDITIONS ARE PASSED ON BY AFFECTED MALES

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will be a carrier, like her mother. There is also a 50% (1 in 2) chance that the daughter will inherit the normal gene. If this happens she will not be a carrier, and will be totally unaffected by the condition. This chance remains the same for every daughter.

If a male who has an X-linked condition has a daughter, he will always pass on the changed gene to her. This is because males only have one X chromosome and they always pass this on to their daughters. All his daughters will therefore be carriers. The daughters will usually not have the condition, but they are at risk of having affected sons.

If a male who has an X-linked condition has a son, his son will never inherit the changed gene on the X chromosome. This is because men always pass on their Y chromosome to their sons (if they passed on their X chromosome they would have a daughter).

### **What if a child is the first person in the family to have the condition?**

Sometimes a child born with an X-linked genetic condition can be the first person to be affected in the family. This may happen because a new gene change has occurred for the first time in the egg or sperm that created the baby.

When this happens, neither parent of that child is a carrier. The parents are very unlikely to have another child affected by the same condition. However the affected child, who now has the changed gene, can pass it on to his or her children.

### **Carrier testing and tests in pregnancy**

A number of options may be available for people who have a family history of an X-linked genetic condition. Carrier testing may be available for females to see if they are carriers of the changed gene. This information may be useful when planning pregnancies.

For some X-linked conditions it is possible to have a test in pregnancy to see if the baby has inherited the condition. This is something you should discuss with your doctor or healthcare professional.

### **Other family members**

If someone in the family has an X-linked condition or is a carrier, you may wish to discuss this with other family members. This gives other female members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. This might be particularly important to family members who already have children, or are likely to have children in the future.

Some people find it difficult to tell other members of the family about a genetic condition. They may be worried about causing anxiety in the family. In some families, people have lost touch with relatives and may feel it is difficult to contact them. Genetic specialists often have a lot of experience with families in these situations and may be able to offer to help you in discussing the situation with other family members.

## Points to remember

- Female carriers have a 50% chance of passing on a changed gene. If a son inherits a changed gene from this mother, then he will be affected by the condition.
- If a daughter inherits a changed gene she will be a carrier like her mother.
- A male who has an X-linked recessive condition will always pass on the changed gene to his daughter, who will then be a carrier. However, if he has an X-linked dominant condition his daughter will be affected. A male will never pass on the changed gene on the X chromosome to his son because his son inherits his Y chromosome (not the X chromosome).
- A changed gene cannot be corrected, it is present for life. However, gene therapy is being developed that may change this in the future.
- A changed gene is not something that can be caught from other people – for example, people with changed genes can still be a blood donor.

## Notes

## Useful links

[bit.ly/rrswanuksupportandinformation](https://bit.ly/rrswanuksupportandinformation)

[bit.ly/whatisageneticdisorder](https://bit.ly/whatisageneticdisorder)

[bit.ly/inheritedgeneticconditions](https://bit.ly/inheritedgeneticconditions)

[bit.ly/yourgenomehome](https://bit.ly/yourgenomehome)

[bit.ly/rrjoinswanuk](https://bit.ly/rrjoinswanuk)

**SWAN UK is a support network run by the charity Genetic Alliance UK, offering support and information to families of children with undiagnosed genetic conditions.**




Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by rare, genetic and undiagnosed conditions. We are an alliance of over 200 patient organisations.



Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK working with the rare disease community and the UK's health departments to effectively implement the UK Strategy for Rare Diseases.

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