

What is “X-linked recessive” inheritance?

Questions and Answers

What are genes?

Genes provide the essential information for the growth and development in each individual. Among others, they determine our height, the colour of our eyes and our risk of developing a particular disorder.

Each cell in our body carries the same set of about **30,000 genes**. They are arranged along so-called chromosomes (Fig. 1). Through a microscope you can see the chromosomes, but not the genes.

How many chromosomes do we have?

Usually, each human body cell contains a complete set of 46 chromosomes. **These 46 chromosomes are made up of 23 pairs**. The pairs numbered 1-22 are referred to as **autosomes**. The chromosomes of the remaining pair are called **sex chromosomes**. They determine whether an individual is female or male. Girls have two X chromosomes and boys have an X and a Y chromosome.

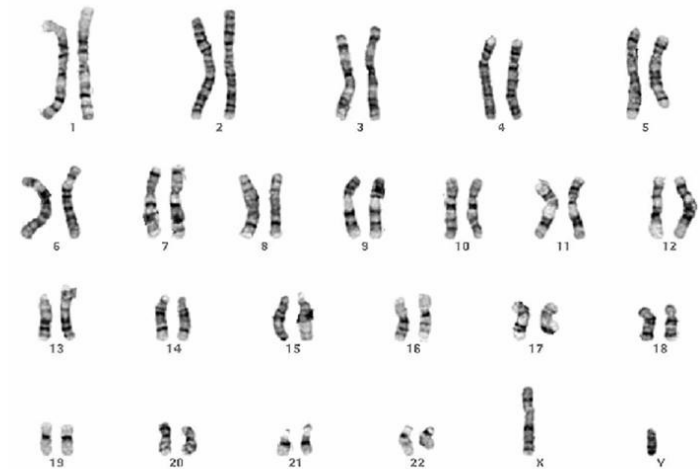


Fig.1 Male set of chromosomes

Of each pair of chromosomes, we inherit one chromosome from our mother and the other from our father. Thus, we have also two copies of each gene, one coming from each of our parents. This is how we inherit characteristic traits from both our parents.

The mother passes on one of her X chromosomes and the father passes on either his X chromosome (in which case the baby will be a girl) or his Y chromosome (the baby will then be a boy).

Each time an egg cell or a sperm is formed, the set of chromosomes is halved from 46 to 23, i.e. each egg or sperm contains chromosomes 1-22 plus a sex chromosome. When an egg cell and a sperm unite (fertilise), the 23 chromosomes of the egg and the 23 chromosomes of the sperm together make up the 46 chromosomes in the new cell. From this fertilised egg, a baby will develop.

What does “X-linked inheritance” mean?

Sometimes, genes carry alterations that cause disorders. Such alterations are called **mutations**. They may give rise to the development of a disease at a certain time in the life of the individual carrying the gene mutation.

A disorder is said to be X-linked if the gene of which a mutated copy exists is located on the X chromosome.

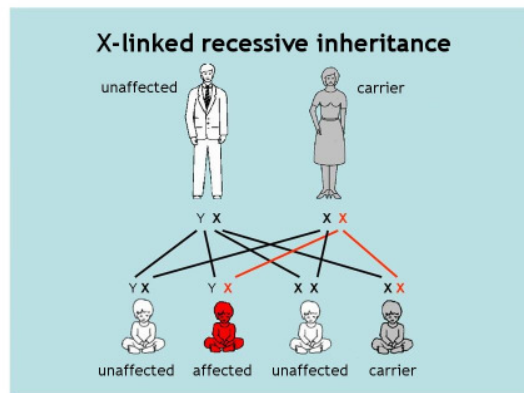


Fig. 2

A girl carrying this mutated gene copy has also an unchanged X chromosome. This compensates for the gene mutation so that, in most cases, the mutation will show no effects on the girl's health. Such a person is called a carrier (heterozygous). A boy carrying the mutated copy of the X chromosome does not have a second X chromosome but has instead a Y chromosome. He will therefore be affected by the condition (Fig. 2).

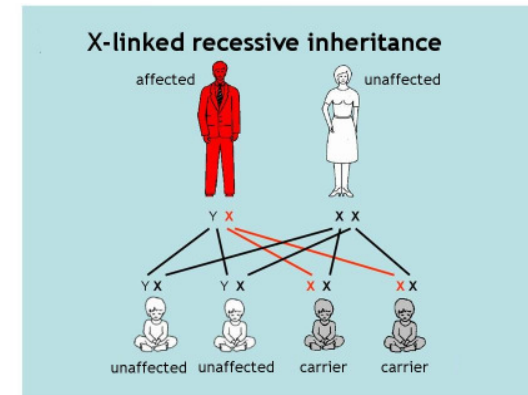


Fig. 3

During the process of fertilisation of an egg by a sperm cell, we have no control over which of the two gene copies coming from the parents is in the egg or the sperm and will be passed on.

If a carrier and her unaffected partner have a son, there is a 1 in 2 (50%) chance that he will be either affected or unaffected. Daughters have a 1 in 2 (50%) chance that they are either carriers of the condition or not. Generally, daughters do not develop the disease. Other probabilities exist in situations where the father is affected and the mother is unaffected and not a carrier of the mutated gene (Fig. 3).

Further information:

You wish to learn more about the inheritance of genetic conditions?

Please contact:

Institute of Human Genetics
 Kerpener Str. 34
 D - 50931 Cologne
 Germany
 Phone: +49-221-478-86811
www.uk-koeln.de/humangenetik