

# What is “autosomal 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.

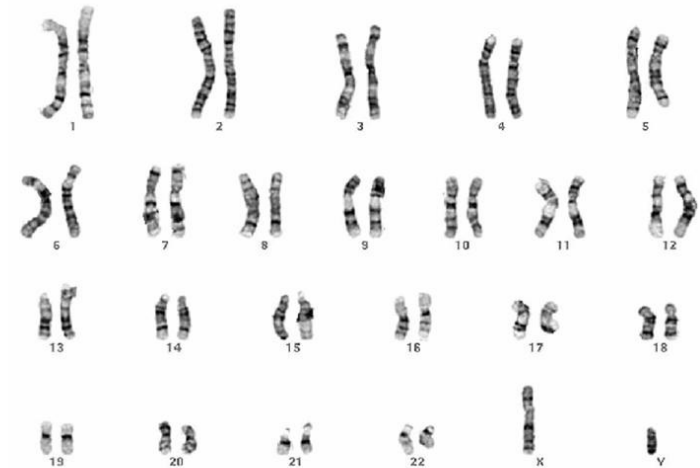


Fig.1 Male set of chromosomes

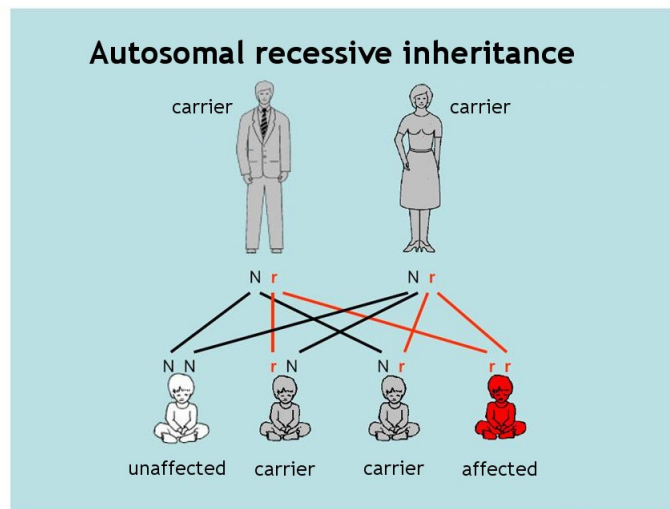
Of each pair of chromosomes, we inherit one chromosome from our mother and the other from our father. Thus, we have also two copies of each gene, one coming from each of our parents. This is how we inherit characteristic traits from both our parents.

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 “recessive 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 recessive if it only becomes manifest in a person if both copies of the same gene are mutated (i.e. the copy coming from the mother and the one from the father).



[http://www.biozentrum.uni-wuerzburg.de/humangenetics/deutsch/HUMANGENETIK\\_1.pdf](http://www.biozentrum.uni-wuerzburg.de/humangenetics/deutsch/HUMANGENETIK_1.pdf)

Fig. 2

People who carry a mutated copy on one chromosome and a correct copy of the same gene on the other chromosome will usually not show any symptoms of the recessive disorder. Such a person is called a carrier (heterozygous) (Fig. 2).

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 you are a carrier and have a baby with a carrier of the same disease, there is a 1 in 4 (25%) chance that the child will be affected by the disease.

The chance is 2 in 4 (50%) that the child will not develop the disease but will be a carrier, just like the parents. The probability that the child will neither be affected nor be a carrier of the condition is 1 in 4 (25%).

In recessive disorders, it is possible to test healthy family members to determine if they are carriers and are thus at risk of having affected children.

Your genetic doctor will be able to explain to you whether any tests are available in your situation.

### Further information:

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

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