

What is “autosomal dominant” 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.

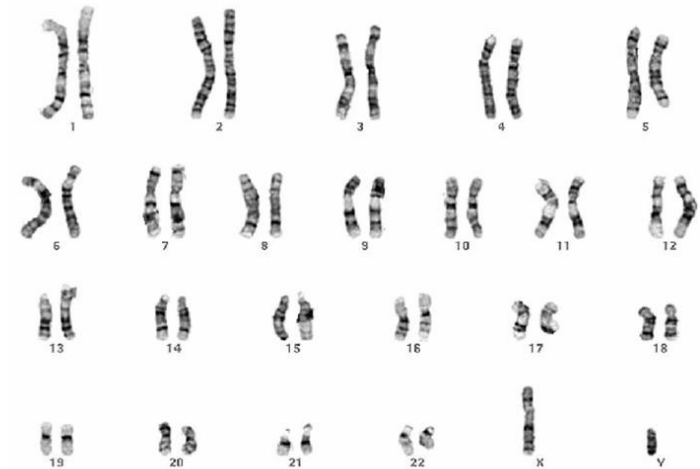


Fig. 1 Male set of chromosomes

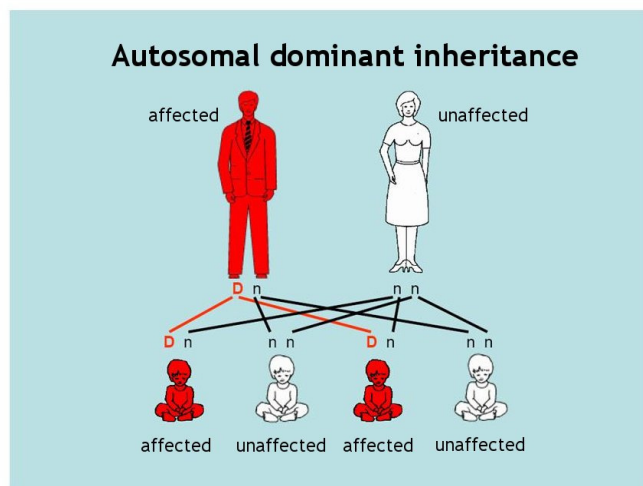
Of each pair of chromosomes, we inherit one chromosome from our mother and the other from our father. Thus, we have also two copies of each gene, one coming from each of our parents. This is how we inherit characteristic traits from both our parents.

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 “dominant 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 dominant if a mutation in one of the two copies of a gene is enough to cause the disease. The unaltered, correct gene copy is then unable to maintain the gene’s full function. The faulty gene copy is **dominant**.



http://www.biozentrum.uni-wuerzburg.de/humangenetics/deutsch/HUMANGENETIK_1.pdf

Fig. 2

If one parent is affected by a disease caused by a dominant mutation, he or she will randomly pass either the correct or the faulty gene copy to their child (Fig. 2).

Thus, the probability is 50% that the child will receive the mutated gene copy and will be affected by the disease. On the other hand,

there is also a 1 in 2 (50%) chance that the child will inherit the correct gene copy. In this case, the child will not develop the disease and will not pass it on to his/her offspring.

How is it that sometimes healthy parents have an affected child?

A gene mutation may occur for the first time in a single egg cell, a sperm or during fertilisation. This is called a new or “spontaneous” mutation. The individual arising from this fertilised egg will be the first affected member of his/her family. If the parents do not carry this alteration, there is only a small risk that further children will also be affected.

The **new mutation** will be present in all body cells of the affected individual and will be passed on to their offspring following the autosomal dominant pattern of inheritance described above. Some disorders affect different people more severely than others. Your genetic doctor will be able to explain to you whether this is also true for your situation.

Further information:

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

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