

Autosomal Dominant Genetic Disorders – An Information Leaflet

A Genetic Disorder is caused by one or more faulty genes

What are genes?

Our bodies are made up of millions of cells and each cell contains a copy of all our genes. Each of us has thousands of genes and they are responsible for all our inherited characteristics, such as eye colour. They act like a set of instructions, controlling how our bodies grow and function. Genes are passed on from parent to child. We each inherit two copies of most genes, one copy from our mother and one copy from our father, forming a pair.

Autosomal Dominant Inheritance

If **one** gene in a pair is faulty, the presence of the other working copy can sometimes compensate for it. This is not the case for all of our genes. For some, both copies must be working properly in order for them to carry out their function. These are called **dominant** genes. The single faulty gene “dominates” the normal gene causing a genetic disorder. If a condition is **autosomal** it means it can affect men **and** women (some conditions affect only one sex, they are called sex linked disorders).

How are dominant genes passed on?

When an affected person has children they may pass on **either** the normal gene **or** the faulty gene to their child. This occurs completely randomly and so there is a 50% or 1 in 2 chance that a child will receive a faulty copy of the gene and may develop the condition. This chance remains the same in each and every pregnancy. There is also a 50% or 1 in 2 chance that a child will receive the correct copy of the gene. In this case, the child will not be affected by the disorder and cannot pass it on to any of his/her children.

The diagram on the following page helps to illustrate this

What if the parents are not affected by the condition?

Sometimes a child born with a dominant genetic condition can be the first person to be affected in a family. All inherited conditions have to start somewhere and occasionally the fault arises in a gene in the egg or sperm of a healthy parent. This fault occurs spontaneously and is called a **new mutation**. This faulty gene can affect just a single egg or sperm and in this case the parents have a very low chance of having another child with the condition. However the affected child who has inherited the faulty gene is at risk of passing it on to the next generation.

Sometimes the faulty gene can arise as egg and sperm cells are being made. When faulty genes arise at this time more than one egg or sperm can be affected. This explains why healthy parents in some rare cases have more than one child with a dominant genetic condition.

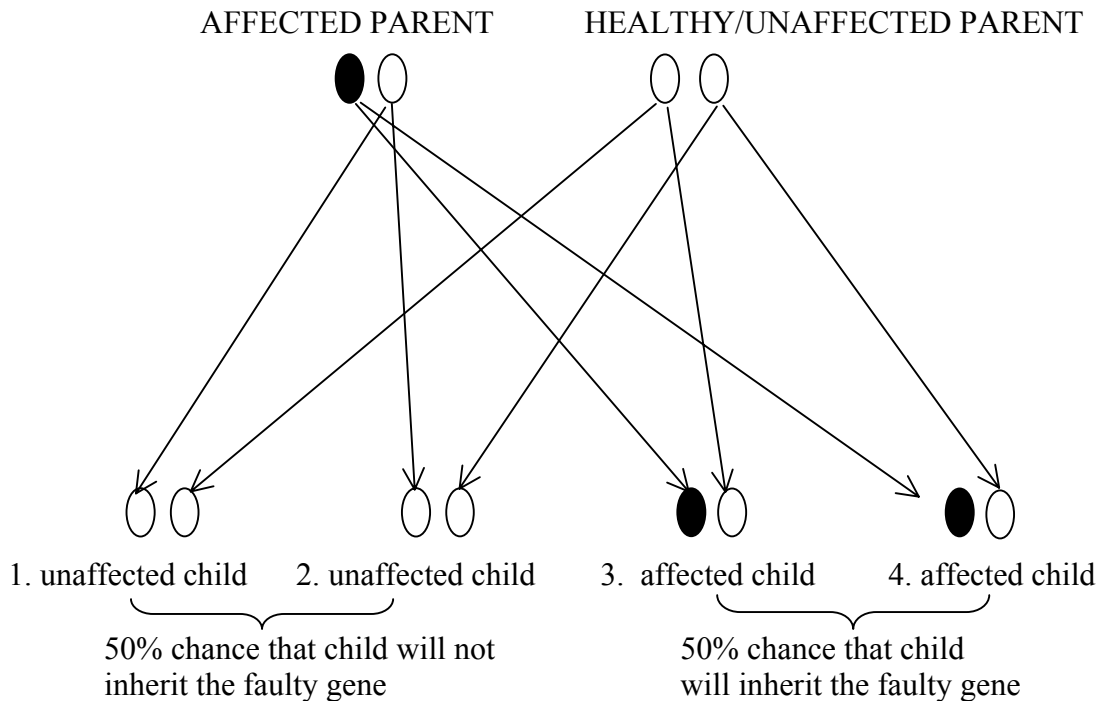
Why does a genetic condition sometimes appear to skip generations?

Some autosomal dominant conditions can be very variable in their effects and not everyone in the family may be affected to the same degree. They do not actually “skip” generations, but in some people the symptoms of the condition are so mild that they appear to be unaffected. In certain conditions that develop after birth, the age that symptoms first appear can also vary greatly between individuals.

How Dominant Genes are passed on from parent to child

- ○ These symbols represent a pair of genes
- This symbol represents a normal/working copy of the gene
- This symbol represents a faulty/alterd copy of the gene

There are FOUR possible ways for the parents to pass on their genes



We cannot include absolutely everything in this leaflet, and each person will have their own particular questions and points they wish to discuss.

If you want further information, please ask to speak to a Doctor or Genetic Nurse Counsellor at:

Northwest Thames Regional Genetics Service (Kennedy Galton Centre), Level 8V, North West London Hospitals NHS Trust, Watford Road, Harrow, Middlesex, HA1 3UJ. Telephone number: 020-8869-2795.

Disclaimer: This leaflet is intended for issue by clinical genetics departments to their patients. The information in this leaflet should not be used for diagnosis. If you think you or your children may be at risk from a genetic disorder in your family, please discuss this with your GP first. Your GP might then refer you to a clinical genetics service. London IDEAS regrets that it cannot enter into discussion of individual cases over the internet, or provide a genetic counseling or diagnostic service.