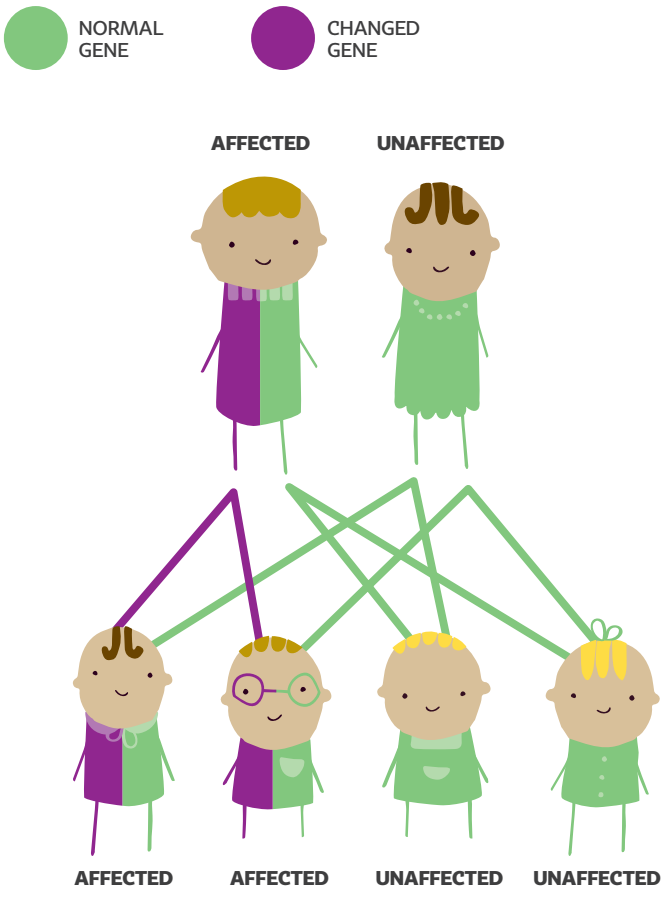
SWAN UK
syndromes without a name

WHAT IS DOMINANT INHERITANCE?

This leaflet gives you information about what dominant inheritance means and how dominant conditions are inherited.

HOW ARE DOMINANT CONDITIONS INHERITED?

How dominant conditions are passed on from parent to child.



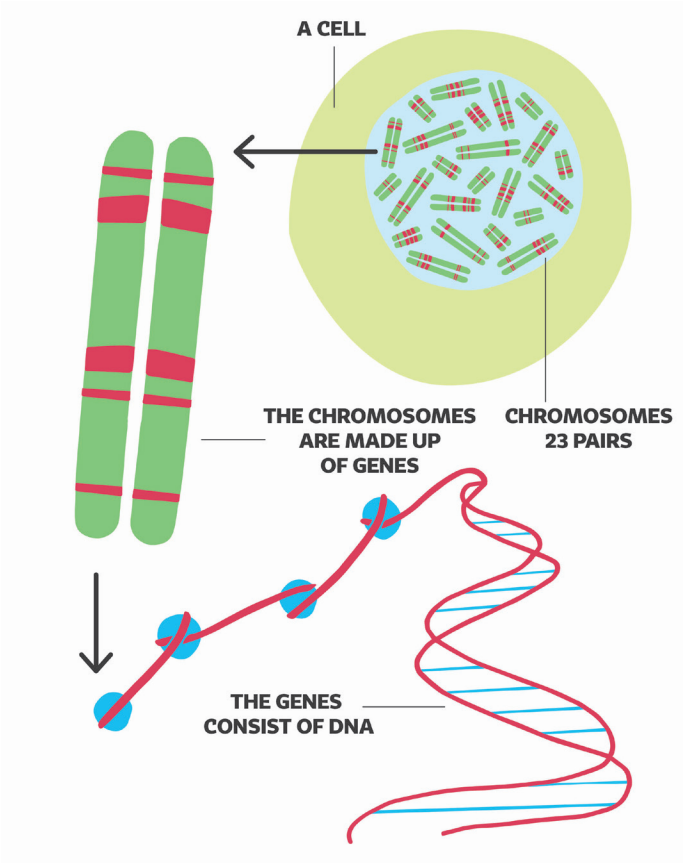
GENETIC ALLIANCE UK

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

Sometimes, there is a change (mutation) 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 to the body.

What is autosomal dominant inheritance?

Some conditions are passed on through the family in a dominant way. This means that if a person inherits one normal copy of a gene, and one changed (mutated) copy, the changed (mutated) gene is dominant over, or overrides, the normal copy. This causes the individual to become affected by the genetic condition. The particular genetic condition that the person is affected by depends on what instructions the changed gene was supposed to give.

Some dominant genetic conditions affect a person from the moment they are born. Others only affect that person during adulthood. These are known as late-onset disorders. Some examples of dominant genetic conditions include autosomal dominant polycystic kidney disease and Huntington's disease.

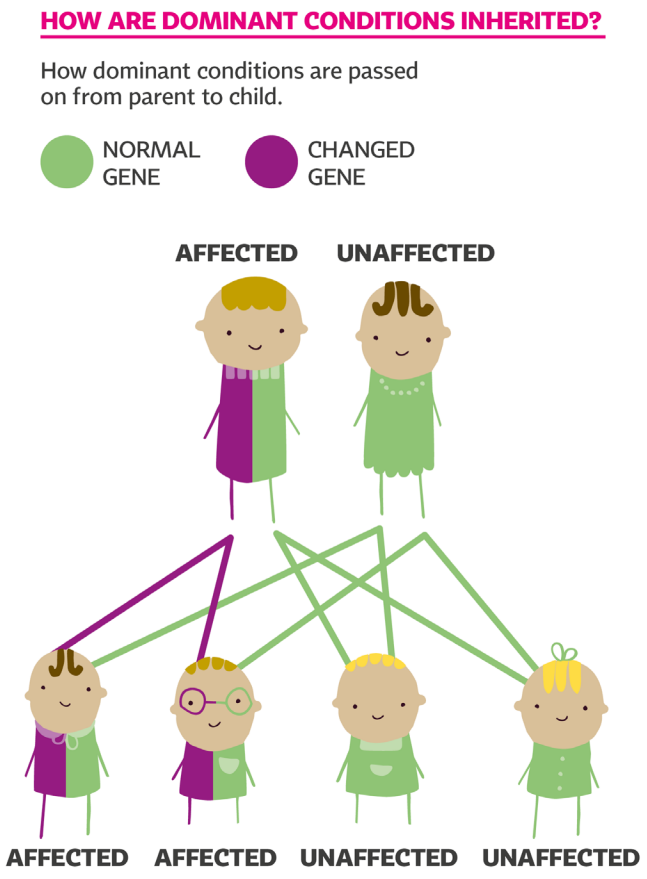
How are dominant genes inherited?

When one parent has a changed gene, they will pass on either their normal gene or their changed gene to their child. Each of their children 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 a child will inherit the normal copy of the gene. If this happens the child will not be affected by the disorder and cannot pass it on to any of his or her children.

These possible outcomes occur randomly. The chance remains the same in every pregnancy and is the same for boys and girls. Very occasionally an affected person will have two copies of the changed gene, so all their children will have the condition.



Why does a genetic condition sometimes appear to miss out a generation?

Some dominant genetic conditions can affect family members very differently. This is called variable expression. The condition does not actually miss out a generation, but some people have such mild symptoms of the condition that they appear to be unaffected. They may not even know that they have the condition.

In conditions which occur later in life (adult-onset conditions, e.g. inherited breast cancer and Huntington's disease) people may have died earlier or of unrelated causes leaving no time for the condition to appear, or the correct diagnosis may never have been given. However, the parents may have passed on the condition to their children.

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

Sometimes a child born with a dominant 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 either the egg or the sperm that went to make that child. When this happens, the parent of that child is not affected. The parents are very unlikely to have another child affected by the same condition, but you should always discuss the risks with your doctor since there are rare occasions when this could occur. However, an affected child, who now has the changed gene, can pass it on to his or her children.

Tests in pregnancy

For some dominant genetic 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.

Points to remember:

- A person only needs to inherit one copy of the changed gene in order to be affected by the condition (50% chance). These outcomes occur randomly. They remain the same in every pregnancy and are the same for boys and girls.
- A changed gene cannot be corrected – it is present for life. However gene therapy is being developed that may be helpful in the future.
- People often feel guilty about a genetic condition which runs in the family. It is important to remember that it is no one's fault and no one has done anything to cause it to happen.

Useful links

bit.ly/rrswanuksupportandinformation

bit.ly/rrxlinkedinheritance

bit.ly/cafamilypatternsofinheritance

bit.ly/rruniquedisorderguides

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.

+44 (0) 20 7831 0883
undiagnosed.org.uk
info@undiagnosed.org.uk

SWANchildrenUK
SWAN_UK
SWANchildrenUK
SWANchildrenUK

Registered charity numbers:
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Registered company number:
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