

This is only a brief guide to dominant inheritance. More information can be obtained from your local regional genetics centre (www.geneticalliance.org.uk/services.htm) or from these addresses:

Genetic Alliance UK

Unit 4D, Leroy House,
436 Essex Rd.,
London, N1 3QP

Telephone: 0207701

Provides information

specific genetic

contact details

organisations.

mail@genetical.org.uk

www.geneticalliance.org.uk

Contact a Family

209-211 City Rd.,
London,

EC

or

56

and

Fri,

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info@cafamily.org.uk

www.cafamily.org.uk

*These contact details may no longer be correct.
Please check them online.
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Orphanet

Free-access website providing information on rare diseases and orphan drugs, and links to support groups across Europe.

www.orpha.net

EuroGentest

Free-access website providing information about genetic testing and links to support groups across Europe.

www.eurogentest.org

Modified from leaflets produced by Guy's and St Thomas' Hospital, London; and the London IDEAS Genetic Knowledge Park, according to their quality standards.

January 2007

This work was supported by EuroGentest, an EU-FP6 supported NoE contract number 512148

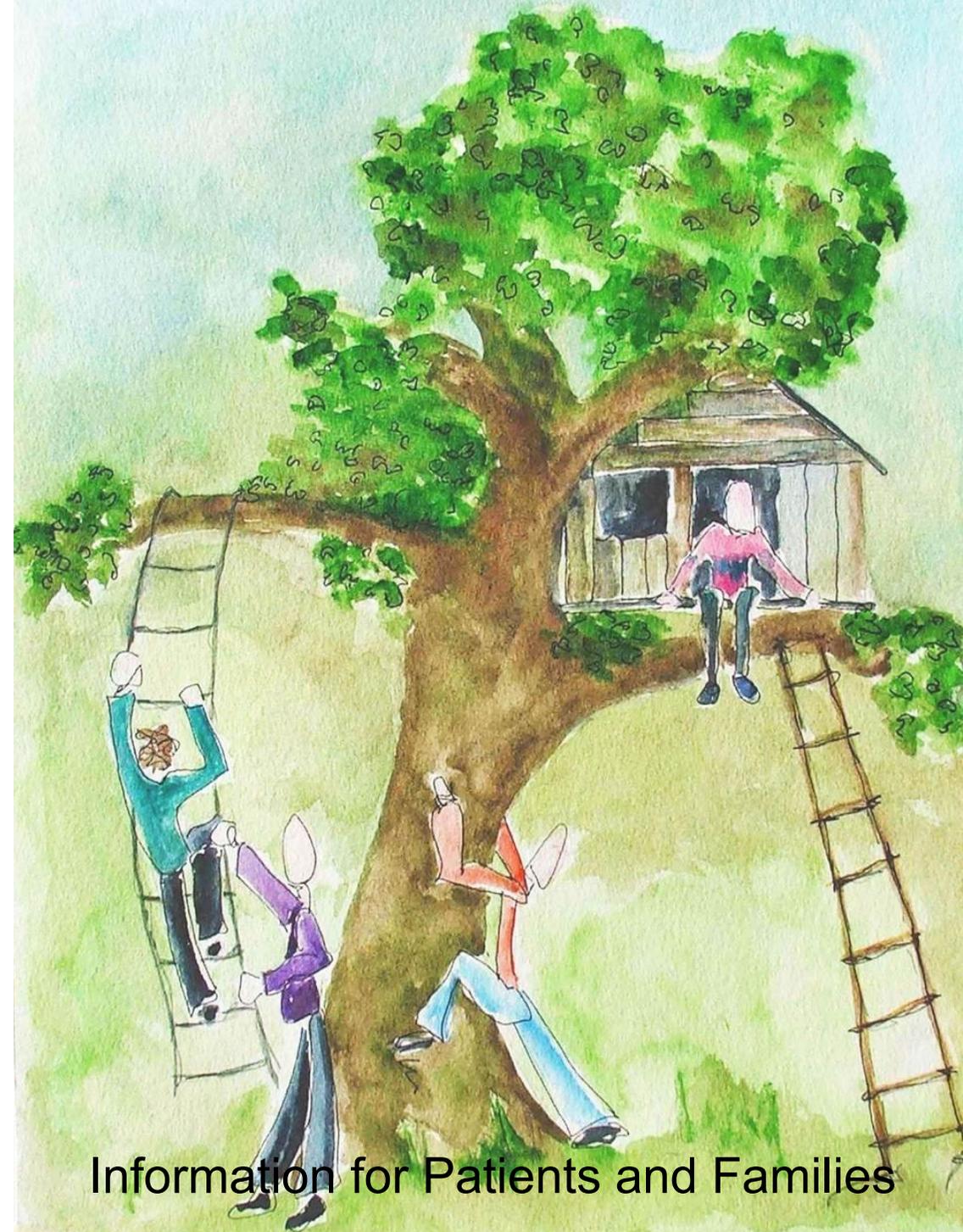
Illustrations by Rebecca J Kent
www.rebeccajkent.com
rebecca@rebeccajkent.com

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Dominant Inheritance



Information for Patients and Families

Dominant Inheritance

The following will give you information about what dominant inheritance means and how dominant conditions are inherited. To understand dominant inheritance, it is first helpful to know about genes and chromosomes.

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 or 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.

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.

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.
- A changed gene is not something that can be caught from other people. They can still be a blood donor, for example.
- 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.



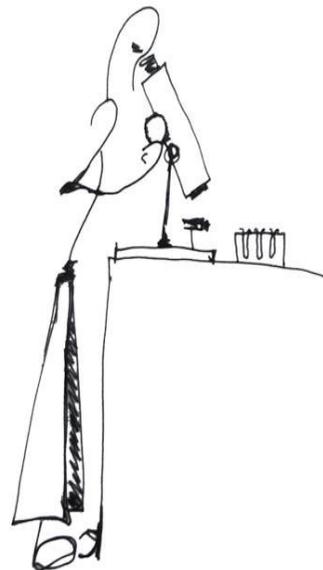
have died earlier of unrelated causes leaving no time for the condition to appear, or the correct diagnosis may never have been given. However, the parents may have passed on the condition to their children.

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

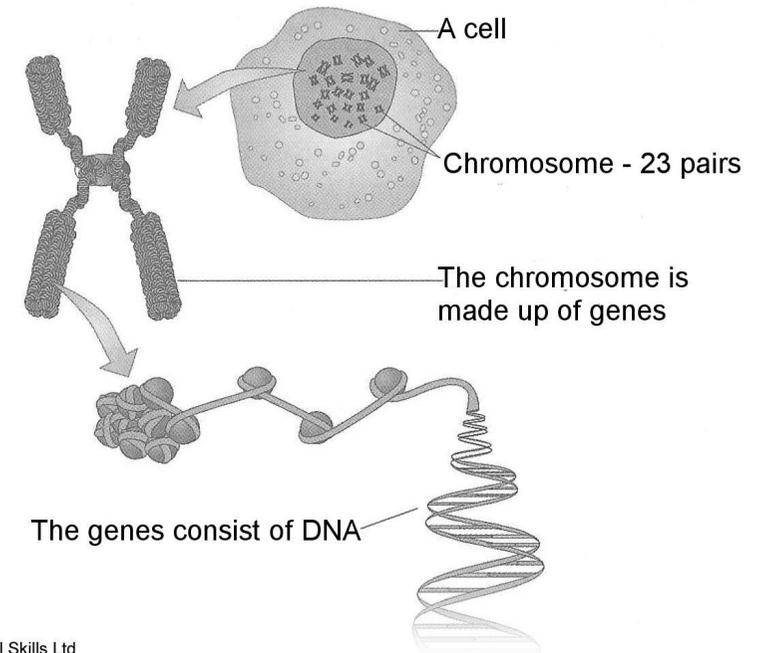
Sometimes a child born with a dominant genetic condition can be the first person to be affected in the family. This may happen because a new gene change has occurred, for the first time, in either the egg or the sperm that went to make that child. When this happens, the parent of that child is not affected. The parents are very unlikely to have another child affected by the same condition, but you should always discuss the risks with your doctor. 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 (more information about these tests are available in the CVS and amniocentesis leaflets). This is something you should discuss with your doctor or healthcare professional.



Picture 1: Genes, chromosomes and DNA



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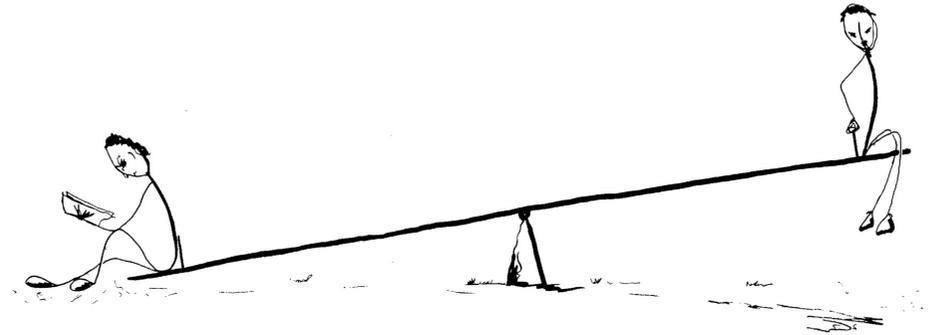
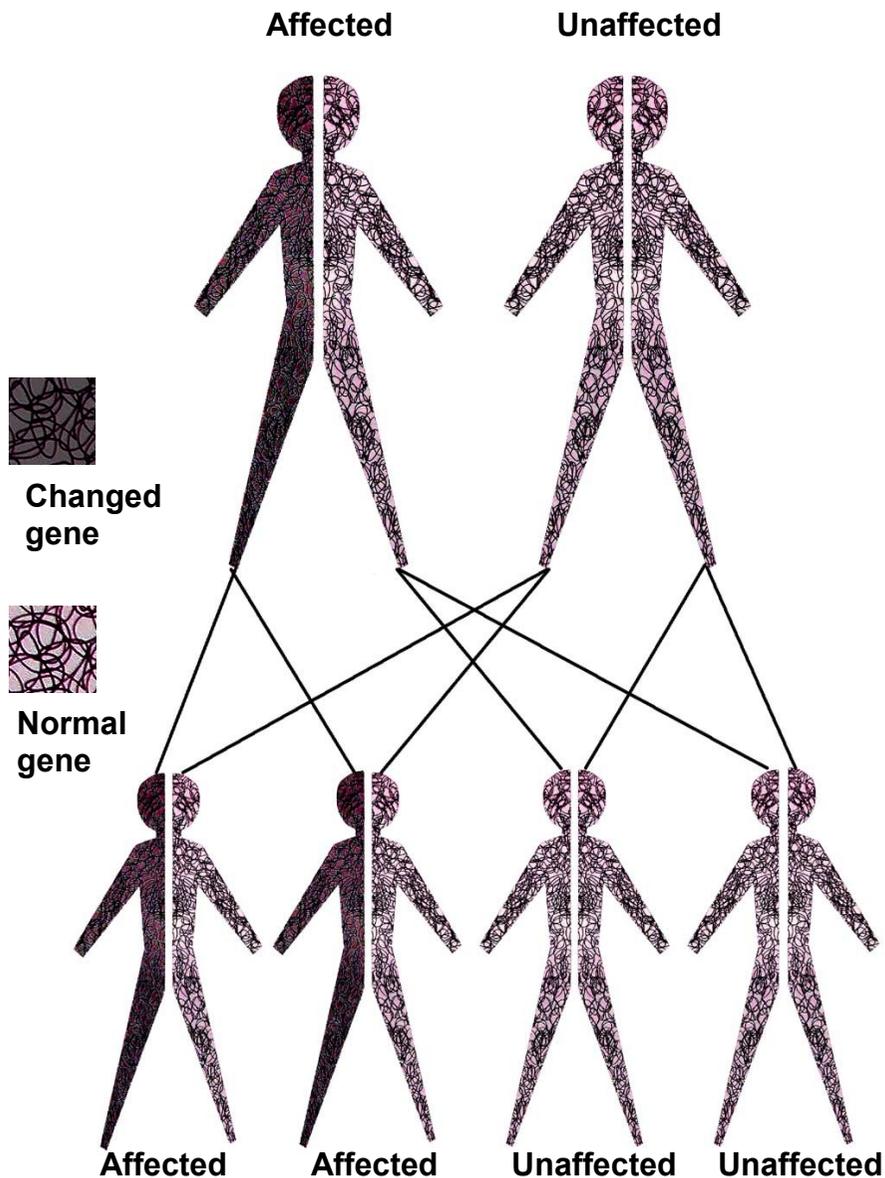
What is autosomal dominant inheritance?

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

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 condition include adult polycystic kidney disease and Huntington's disease.

How are dominant conditions inherited?

Picture 2: How dominant conditions are passed on from parent to child



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.

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