This is only a brief guide to recessive 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: 0207704 3141
Provides information about specific genetic conditions and contact details of support organisations.
mail@geneticalliance.org.uk
www.geneticalliance.org.uk

Contact a Family
209-211 City Rd.,
London,
EC1V 1JN
Helpline 0808 808 3555 or
Textphone 0808 808 3556
(Freephone for parents and families, 10am-4pm Mon-Fri, 4.30pm-7.30pm Mon)
info@cafamily.org.uk
www.cafamily.org.uk

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

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

EuroGentest

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Recessive Inheritance

Information for Patients and Families

Recessive Inheritance

The following will give you information about what recessive inheritance means and how recessive conditions are inherited. To understand recessive 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 contains 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.

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. If this occurs in only one recessive gene, and the person has another normal copy, this will not usually cause a genetic condition.

Points to remember

- A person must inherit two copies of a changed gene, one from each parent, in order to be affected by the condition (25% chance). If a person inherits only one changed gene then they will be a carrier (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.

Carrier Testing and Tests in Pregnancy

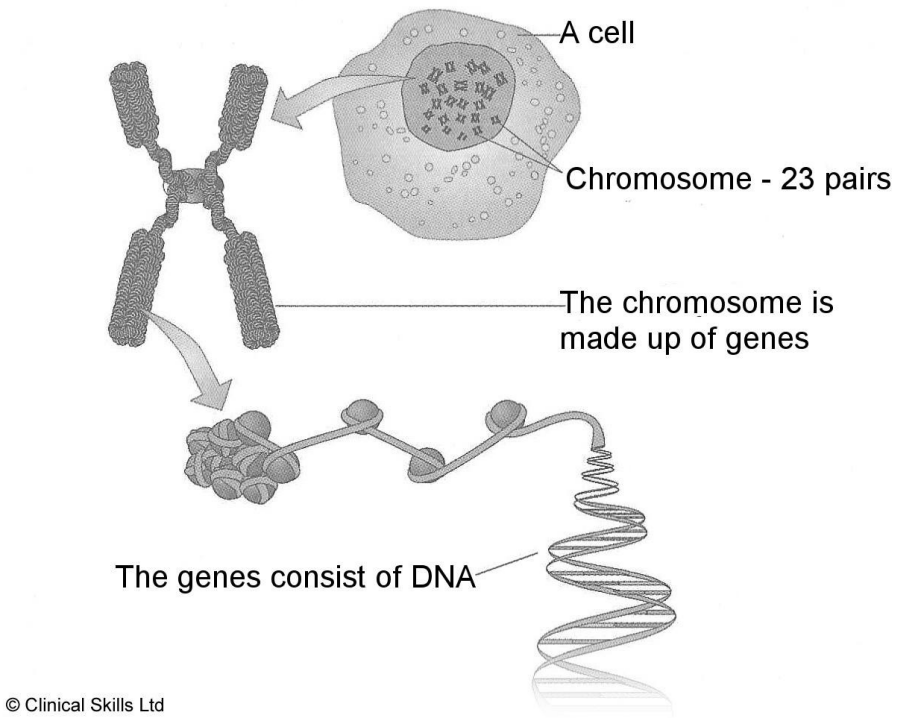
A number of options may be available for people who have a family history of a recessive genetic condition. Carrier testing may be available to see if the couple are both carriers of the changed gene. This information may be useful when planning pregnancies. For some recessive conditions, it is possible to have a test in pregnancy to see if the baby has inherited the condition (more information about these test are available in the CVS and amniocentesis leaflets). This is something you should discuss with your doctor or healthcare professional.

Other Family Members

If someone in the family has a recessive condition or is a carrier, you may wish to discuss this with other family members. This gives other family members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. This might be particularly important to family members who already have children, or are likely to have children in the future.

Some people find it difficult to tell other members of the family about a genetic condition. They may be worried about causing anxiety in the family. In some families, people have lost touch with relatives and may feel it is difficult to contact them. Genetic specialists often have a lot of experience with families in these situations and may be able to offer you help in discussing the situation with other family members.

Picture 1: Genes, chromosomes and DNA

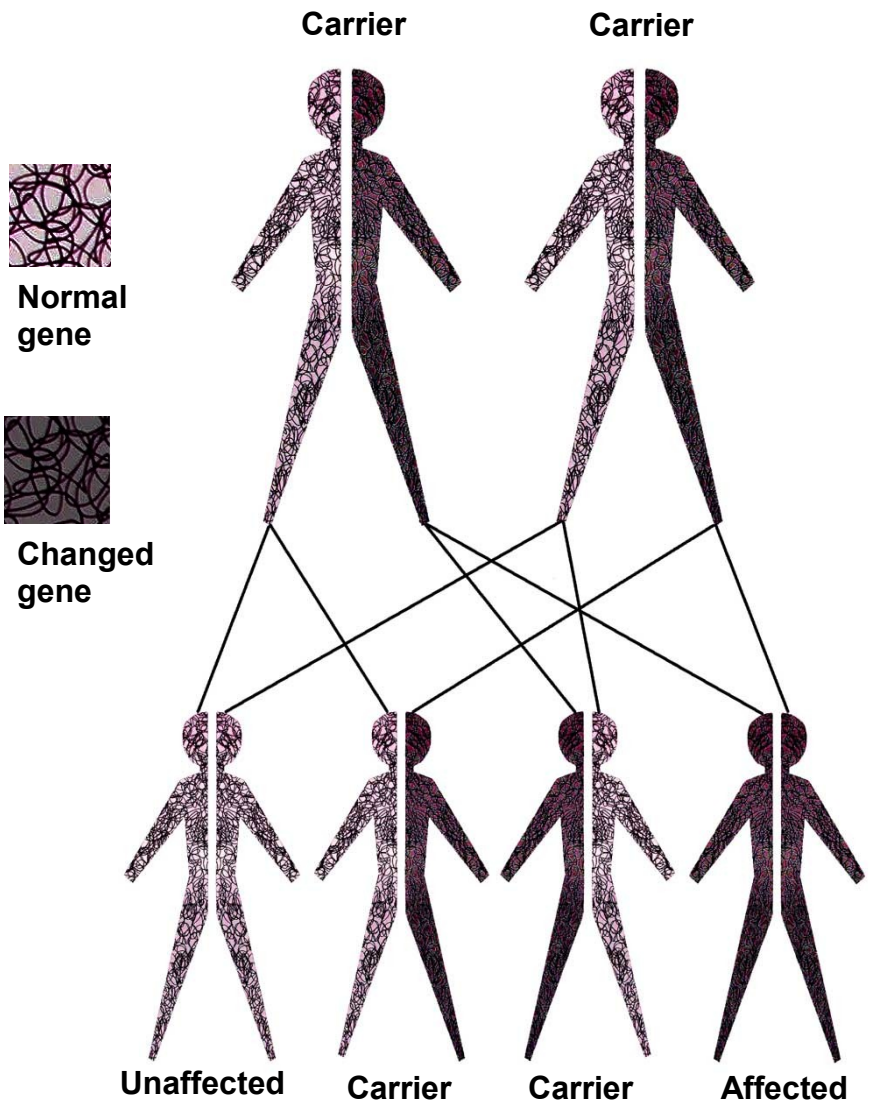


What is autosomal recessive inheritance?

Some conditions are inherited as recessive conditions. This means that a person must inherit two changed copies of the same gene (one changed copy from each parent) in order to have the condition. If a person inherits one changed copy and one normal copy, then in most cases that person will be a healthy carrier because the normal copy compensates for the changed copy. Being a carrier means that you do not have the condition, but carry a changed copy of the gene on one of a pair of genes. Examples of autosomal recessive conditions include cystic fibrosis and sickle-cell anaemia.

How are recessive conditions inherited?

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



If both partners are carriers of the same changed gene, they may pass on either their normal gene or their changed gene to their child. This occurs randomly.

Each child of parents who both carry the same changed gene therefore has a 25% (1 in 4) chance of inheriting a changed gene from both parents and being affected by the condition.

This also means that there is a 75% (3 in 4) chance that a child will not be affected by the condition. This chance remains the same in every pregnancy and is the same for boys or girls.

There is also a 50% (2 in 4) chance that the child will inherit just one copy of the changed gene from a parent. If this happens, then they will be healthy carriers like their parents.

Lastly, there is a 25% (1 in 4) chance that the child will inherit both normal copies of the gene. In this case the child will not have the condition, and will not be a carrier.

These possible outcomes occur randomly. The chance remains the same in every pregnancy and is the same for boys and girls.