

This is only a brief guide to chromosome changes. More information can be obtained from your local regional genetics centre (www.gig.org.uk/services.htm) and from these addresses:

**Unique -
The Rare Chromosome
Disorder Support Group**
PO Box 2189,
Caterham,
Surrey
CR3 5GN
Telephone: 01883 330766
Email: info@rarechromo.org
Web: www.rarechromo.org

The Genetic Interest Group
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.
Email: mail@gig.org.uk
Web: www.gig.org.uk

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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)
Email: info@cafamily.org.uk
Web: www.cafamily.org.uk

**Antenatal Results and
Choices (ARC)**
Tel: 020 7631 0285
Email: info@arc-uk.org
Web: www.arc-uk.org

EuroGentest and Orphanet
Free-access websites
providing information about
genetic testing and links to
support groups across Europe.
Web: www.eurogentest.org
Web: www.orpha.net

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Chromosome Changes



Information for Patients and Families

Chromosome Changes

The following information discusses what chromosome changes are, how they are inherited and when they might cause problems. This information is designed to be used alongside the discussions you have with your genetic specialist.

In order to understand chromosome changes, it is helpful to know about genes and chromosomes.

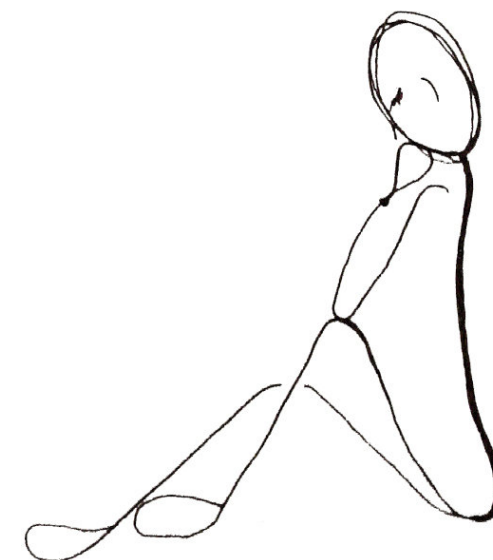
What are genes and chromosomes?

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. We have thousands of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are responsible for many of our characteristics, such as our eye colour, blood type or height.

Genes are carried on thread-like structures called chromosomes. Usually, we have 46 chromosomes in most cells. We inherit our chromosomes from our parents, 23 from our mother and 23 from our father, so we have two sets of 23 chromosomes, or 23 'pairs'. Because the chromosomes are made up of genes we therefore inherit two copies of most genes, one copy from each parent. This is the reason why we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

Points to remember

- A rearrangement is either inherited from a parent or happens around the time of conception.
- A rearrangement cannot be corrected – it is present for life.
- A rearrangement is not something that can be caught from other people. Therefore a translocation carrier can still be a blood donor, for example.
- People often feel guilty about something like a chromosome rearrangement 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.
- The vast majority of carriers of a balanced rearrangement are able to have healthy children.



Frequently a child can be born with a rearrangement although both parents' chromosomes are normal. This is called a "*de novo*" (from Latin) or new rearrangement. In this case the chance that the parents will have another child with the same rearrangement is usually very low.

Tests for chromosome rearrangements

Genetic testing is available to find out whether a person carries a rearrangement. A simple blood test is done, and cells from the blood are examined in a laboratory to look at the arrangement of the chromosomes. This is called a karyotype test. It is also possible to do a test during pregnancy to check the baby's chromosome structure. This is called prenatal diagnosis and is something you may wish to discuss with the genetic specialist (more information about these tests are available in the CVS and amniocentesis leaflets).

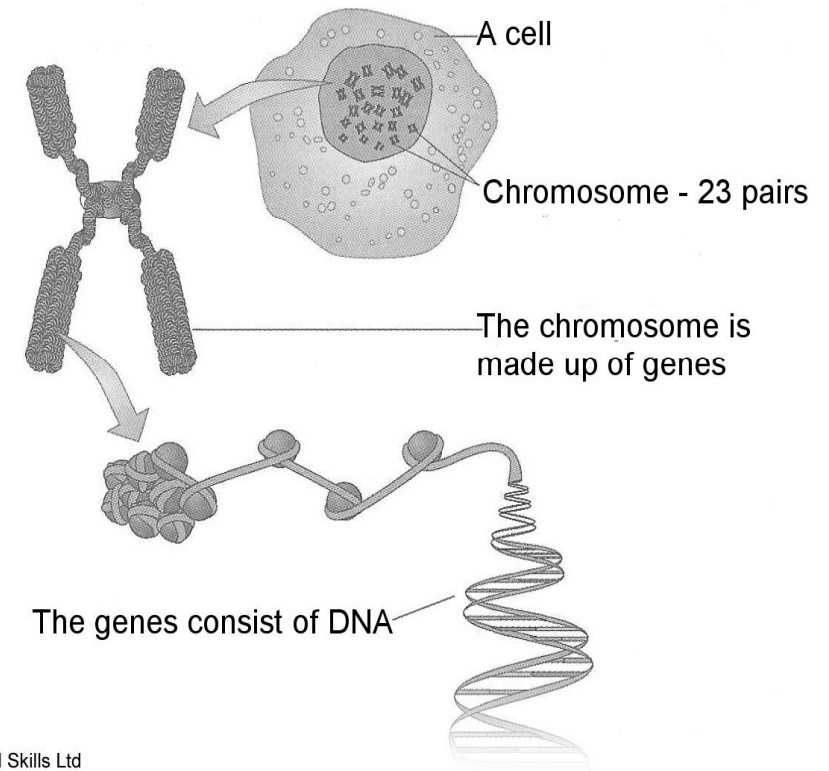
What about other family members

If a rearrangement is found in a family member, 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 also carry the rearrangement, if they wish. This might be particularly important to family members who already have children, or are likely to have children in the future.

If they do **not** carry the rearrangement then they **cannot pass it on** to their children. If they do carry it they too could be offered a test during pregnancy to check the baby's chromosomes.

Some people find it difficult to tell other members of the family about the rearrangement. 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

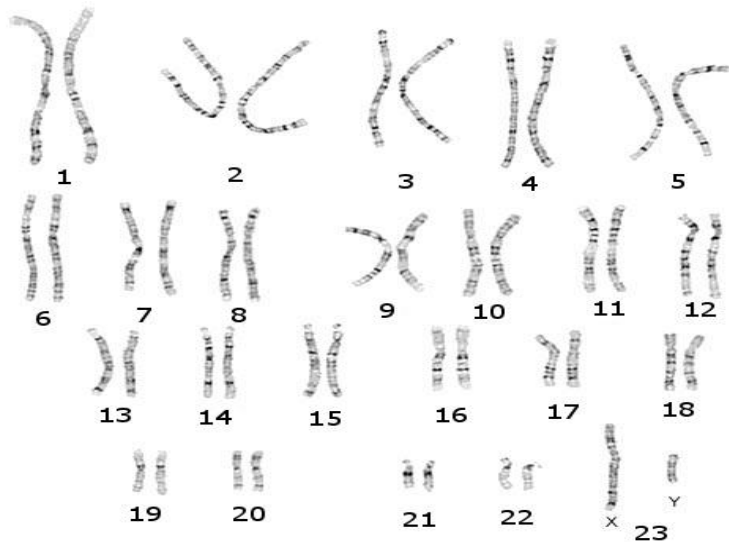


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The chromosomes (see **Picture 2**) numbered 1 to 22 look the same in males and females. These are called the autosomes. Pair number 23 is different in males and females and they are called the sex chromosomes. There are two kinds of sex chromosome, one called the X chromosome and one called the Y chromosome. Females normally have two X chromosomes (XX). A female inherits one X chromosome from her mother and one X chromosome from her father.

Males normally have an X and a Y chromosome (XY). A male inherits an X chromosome from his mother and a Y chromosome from his father. The picture below therefore shows the chromosomes of a male as the last pair of chromosomes are (XY).

Picture 2: 23 pairs of chromosomes arranged according to size; chromosome 1 is the largest. The last two chromosomes are sex chromosomes.

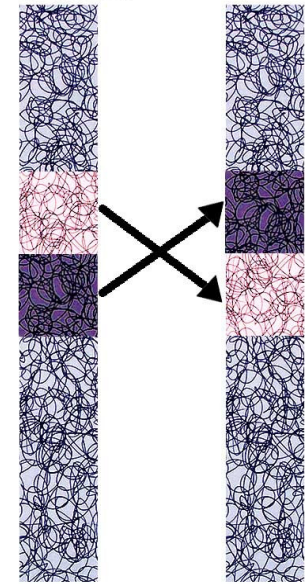


Chromosome Changes

It is important that we have the correct balance of chromosome material. This is because the genes, that instruct the cells in our body, are found on the chromosomes. Any change in the number, size or structure of our chromosomes can mean a change in the amount or arrangement of genetic information. A change in the amount or arrangement of genetic information may result in learning disability, developmental delay and health problems in a child.

Inversions

Chromosome inversion means that part of a chromosome has turned so that the sequence of genes in the chromosome is partly reversed. In the majority of cases this does not cause any health problems to the person carrying the inversion.



If a parent has an unusual chromosome rearrangement, will he or she always pass it on?

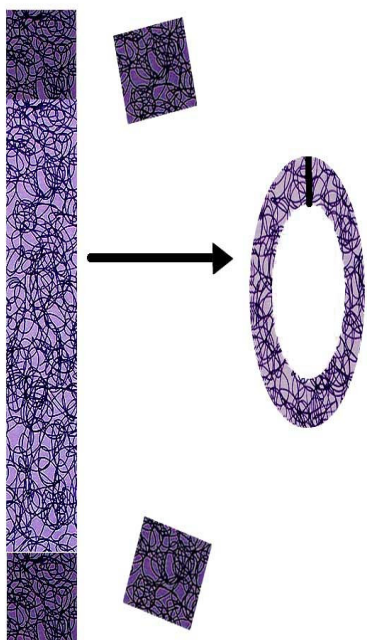
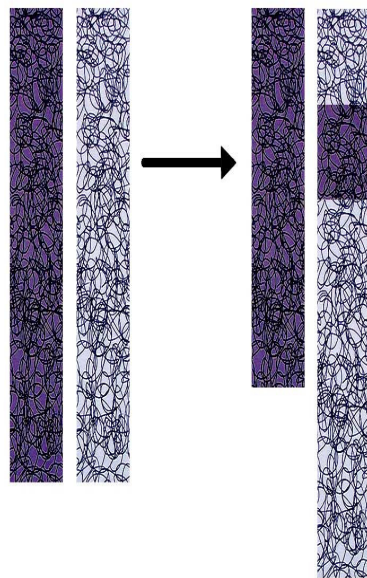
Not necessarily, there are several possibilities for each pregnancy:

- The child may inherit an entirely normal chromosome arrangement.
- The child may inherit the same chromosome rearrangement as the parent.
- The child may be born with learning disability, developmental delay and health problems.
- The pregnancy ends in miscarriage.

Therefore it is usually possible for a person who carries a chromosome rearrangement to have healthy children, and many do. As each rearrangement is unique, carriers would have to discuss their particular situation with a genetic specialist.

Insertions

Chromosome insertion means that material from a chromosome has been inserted into an unusual position in the same or another chromosome. If there is **no** additional or missing chromosome material, that person is usually healthy. However if there **is** additional or missing chromosome material, then that person may have learning disability, developmental delay and health problems.



Rings

The term chromosome ring means that the ends of a chromosome have joined together in a 'ring' shape. This usually happens when the two ends of the same chromosome are deleted. The remaining ends of the chromosome are 'sticky' and join together to make a ring shape. The effect this has on the person usually depends on how much chromosome material, and therefore 'information', was deleted before the chromosome formed a 'ring'.

Chromosome changes may be inherited from a parent. More commonly, chromosome changes happen either when the egg or sperm cells are made, or around the time of conception. These changes occur without us being able to control them.

There are two main types of chromosome changes that can occur.

Changes in chromosome number. This is when there are more or fewer copies of a particular chromosome than usual.

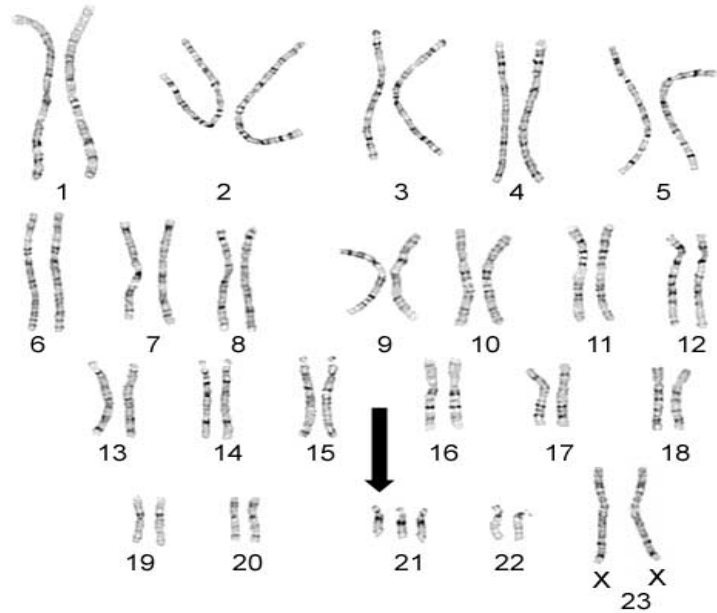
Changes in chromosome structure. This is when the material in an individual chromosome is disrupted or rearranged in some way. This may involve the addition or loss of chromosome material. In this leaflet we will be looking at chromosome **deletions**, chromosome **duplications**, chromosome **insertions** and chromosome **inversions** and **rings**. If you would like information about chromosome **translocations**, please look at the Chromosome Translocations leaflet.

Changes in chromosome number

Usually, every cell in a person's body contains 46 chromosomes. Sometimes however a baby is born with either too many or too few chromosomes. The baby therefore has too few or too many genes or instructions.

One of the most common examples of a genetic condition caused because of an extra chromosome is Down syndrome. People with this condition have 47 chromosomes in their cells instead of 46. This is because there are three copies of chromosome number 21 instead of the usual two.

Picture 3: Chromosomes from a girl (last pair of chromosomes are XX) with Down syndrome. There are three copies of chromosome 21 instead of the usual two.



Changes in chromosome structure

Changes in chromosome structure happen when the material in an individual chromosome is broken and rearranged in some way. This may involve the addition or loss of chromosome material. This may happen in a number of ways which are discussed below.

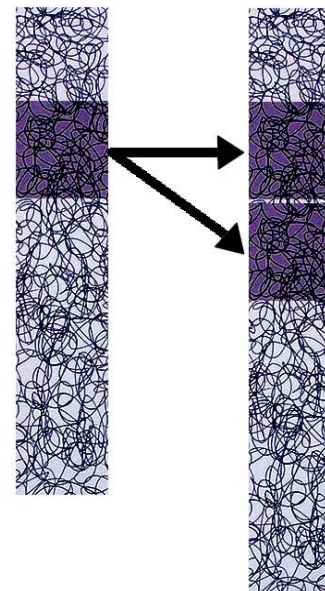
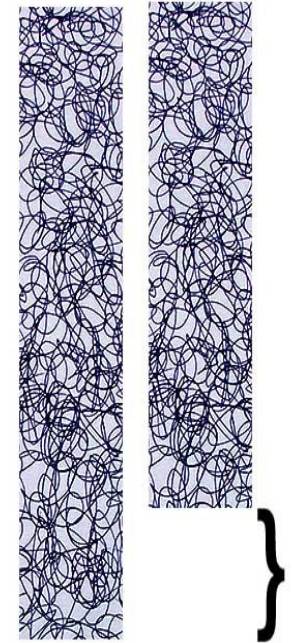
Changes in chromosome structure may be very subtle and hard to detect by scientists in the laboratory. Even when the change in structure is found, it is often hard to predict what effect the change will have on an individual child. This can be frustrating for parents who are keen to have as much information about their child's future as possible.

Translocations

If you would like information about translocations, please look at the Chromosome Translocations leaflet.

Deletions

The term chromosome deletion means that part of a chromosome has been lost or deleted. A deletion can happen on any chromosome, and along any part of the chromosome. The deletion can be any size. If the material (genes) that has been deleted contains important instructions for the body, that person may have learning disability, developmental delay and health problems. The seriousness of these depends on how much of the chromosome has been deleted, and where the deletion is.



Duplications

The term chromosome duplication means that the chromosome has duplicated part of itself, so that there is too much chromosome material present. This additional chromosome material may mean there are too many instructions for the body to process, and this may result in learning disability, developmental delay and health problems in a child.