

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

### Unique- The Rare Chromosome Disorder Support Group

Telephone: 01883 330766  
[info@rarechromo.org](mailto:info@rarechromo.org)  
[www.rarechromo.org](http://www.rarechromo.org)

### Genetic Alliance UK

Unit 4D, Leroy House  
 436 Essex Rd  
 London  
 Telephone: 020 7631 0285  
 Provision of information from Mon-Fri, 9am-5pm (London)  
[www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)  
 specific contact details for each of our member organisations  
[mail@geneticalliance.org.uk](mailto:mail@geneticalliance.org.uk)  
[www.geneticalliance.org.uk](http://www.geneticalliance.org.uk)

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

### Contact a Family

209-211 City Rd.,  
 London, EC1V 1JN  
 Helpline 020 7631 8083 3555 or  
 Telephone 020 7631 8083 3556  
 Provision of information from Mon-Fri, 9am-5pm (London)  
[www.family.org.uk](http://www.family.org.uk)  
[info@family.org.uk](mailto:info@family.org.uk)

### Antenatal Results and Choices (ARC)

Tel: 020 7631 0285  
[info@arc-uk.org](mailto:info@arc-uk.org)  
[www.arc-uk.org](http://www.arc-uk.org)

### EuroGentest and Orphanet

Free-access websites providing information about genetic testing and links to support groups across Europe.  
[www.eurogentest.org](http://www.eurogentest.org)  
[www.orpha.net](http://www.orpha.net)

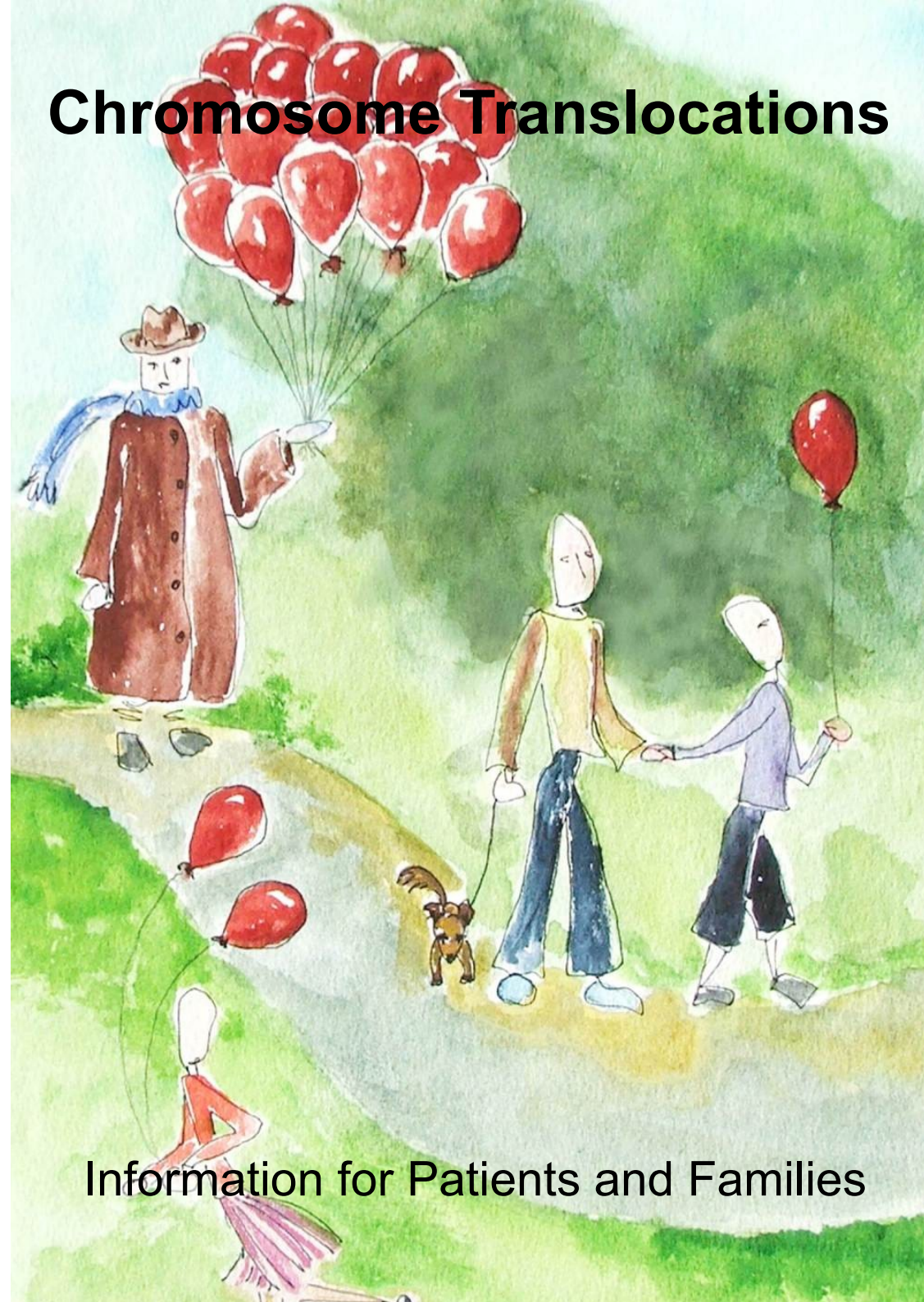
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[www.rebeccajkent.com](http://www.rebeccajkent.com)  
[rebecca@rebeccajkent.com](mailto:rebecca@rebeccajkent.com)

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# Chromosome Translocations



Information for Patients and Families

## Chromosome Translocations

The following information discusses what chromosome translocations 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.

### What is a chromosome translocation?

To understand what a chromosome translocation is, it is helpful to know about genes and chromosomes.

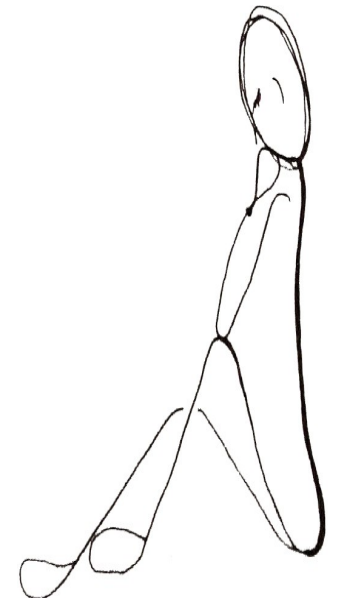
### What are genes and chromosomes?

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. 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.



## Points to remember

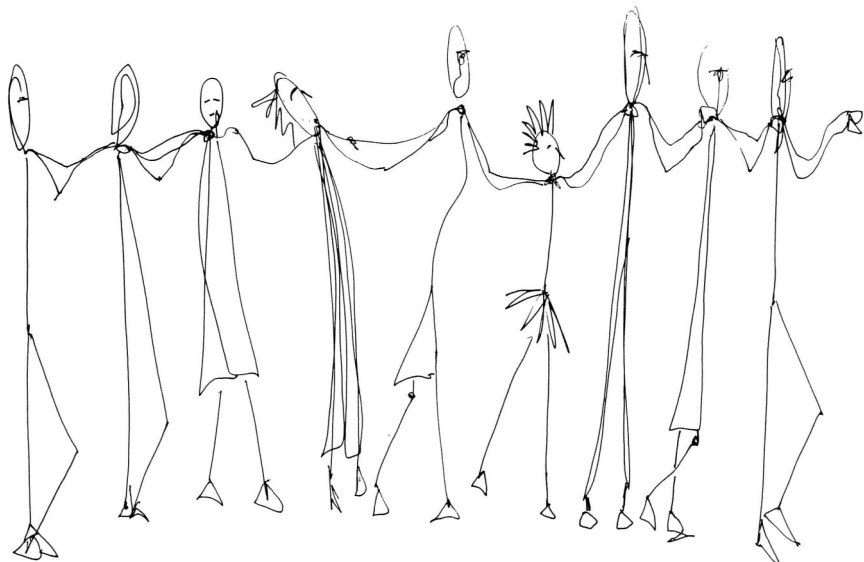
- People who carry a **balanced translocation** are usually healthy. The only time that a problem may arise is when that person tries to have children.
- A translocation is either inherited from a parent or happens around the time of conception.
- A translocation cannot be corrected – it is present for life.
- A translocation 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 balanced translocation 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.



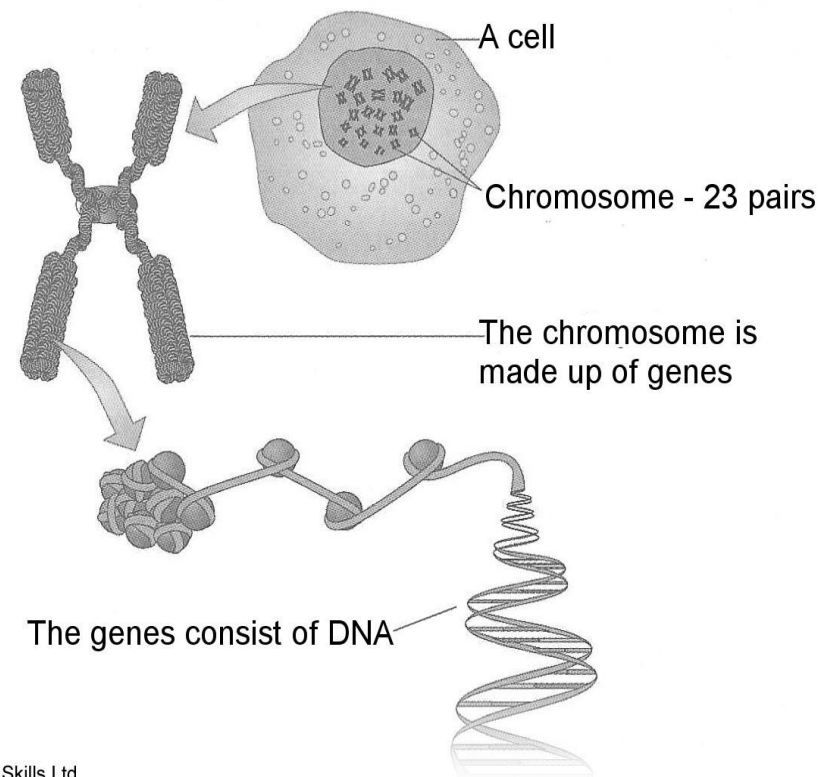
## What about other family members?

If a translocation is found, that person 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 translocation, 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 a translocation then they **cannot pass it on** to their children, but if they do carry a translocation 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 translocation. 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.



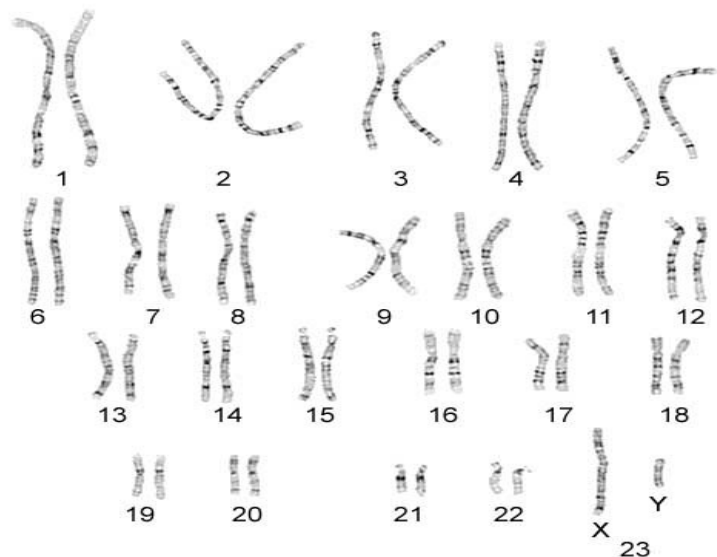
## Picture 1: Genes, chromosomes and DNA



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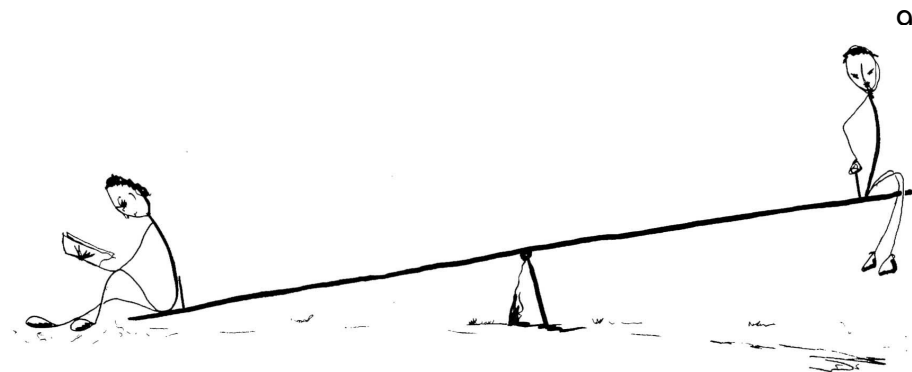
Genes are located on small thread-like structures called chromosomes. Usually we have 46 chromosomes in most cells. One set of 23 chromosomes we inherit from our mother and one set of 23 chromosomes we inherit from our father. So we have two sets of 23 chromosomes, or 23 pairs.

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



Pairs number 1 to 22 look the same in males and females. These are called the autosomes. However the 23<sup>rd</sup> pair, known as the sex chromosomes, are different in males and females. There are two sex chromosomes, the 'X' chromosome and the 'Y' chromosome. Females have two X chromosomes (XX), whilst males have an X chromosome and a Y chromosome (XY). A female inherits one X chromosome from her mother and one X chromosome from her father. A male inherits an X chromosome from his mother and a Y chromosome from his father. The picture above therefore shows the chromosomes of a male as the last pair of chromosomes are XY.

It is important that we have the correct amount of chromosome material, as the genes (that instruct the cells in our body) are found on the chromosomes. Having some part of a chromosome missing, or having an extra part of a chromosome, can result in learning difficulties, developmental delay and health problems in a child.



Therefore it is quite possible for a person who carries a balanced translocation to have healthy children, and many do. However, the risk that a carrier of a balanced translocation will have a child with some degree of disability is higher than average, although the severity of the disability depends on the exact type of translocation.

### Tests for chromosome translocations



Genetic testing is available to find out whether a person carries a translocation. 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 find out whether a baby has a chromosome translocation. 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).

## Unbalanced translocations

If either parent carries a balanced translocation, it is possible that their child may inherit an **unbalanced translocation** in which there is an extra piece of one chromosome and/or a missing piece of another chromosome.

Frequently a child can be born with a translocation although both parents' chromosomes are normal. This is called a "*de novo*" (from Latin) or new rearrangement. In this case the parents are unlikely to have another child with a translocation.

A child who has an unbalanced translocation may have learning disability, developmental delay and health problems. The seriousness of the disability depends on exactly which parts of which chromosomes are involved and how much missing or extra chromosome material there is. This is because some parts of the chromosome are more important than other parts.

### If a parent has a balanced translocation will he or she always pass it on?

Not necessarily, there are several possibilities for each pregnancy:

- The child may inherit entirely normal chromosomes.
- The child may inherit the same balanced translocation as the parent. In most cases the child will not have any problems as a result of the translocation.
- The child may inherit an unbalanced translocation, and may be born with some degree of developmental delay, learning disability and health problems.
- The pregnancy ends in miscarriage.

## What is a translocation?

A translocation means that there is an unusual arrangement of the chromosomes. This can happen because:

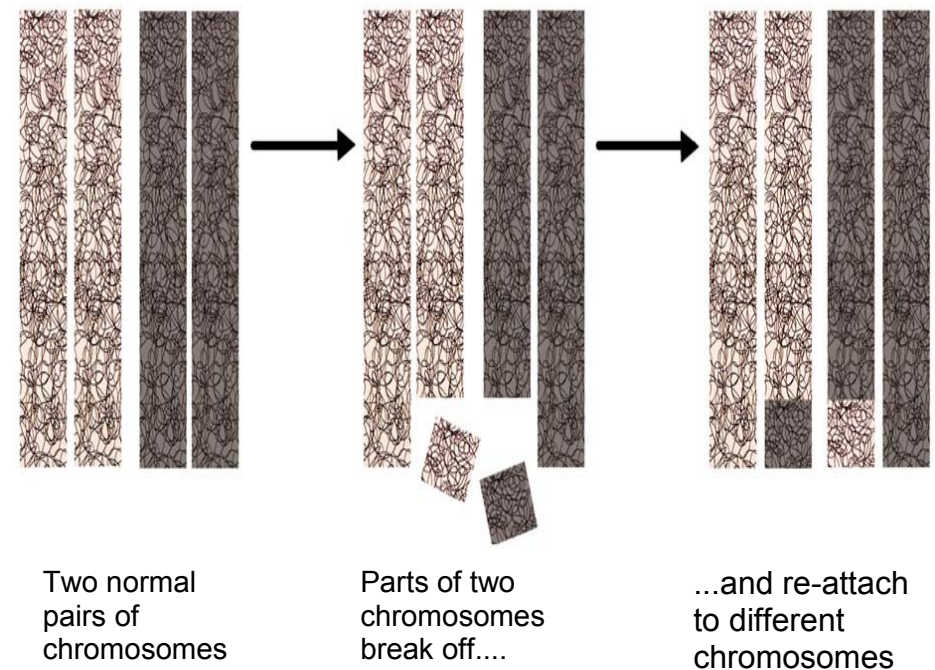
- a) a change has arisen during the making of the egg or the sperm or around the time of conception
- b) an altered chromosome arrangement has been inherited from either the mother or the father

There are two main types of translocations: a **RECIPROCAL** translocation and a **ROBERTSONIAN** translocation.

### Reciprocal translocations

A reciprocal translocation occurs when two fragments break off from two different chromosomes and swap places. This can be seen in Picture 3.

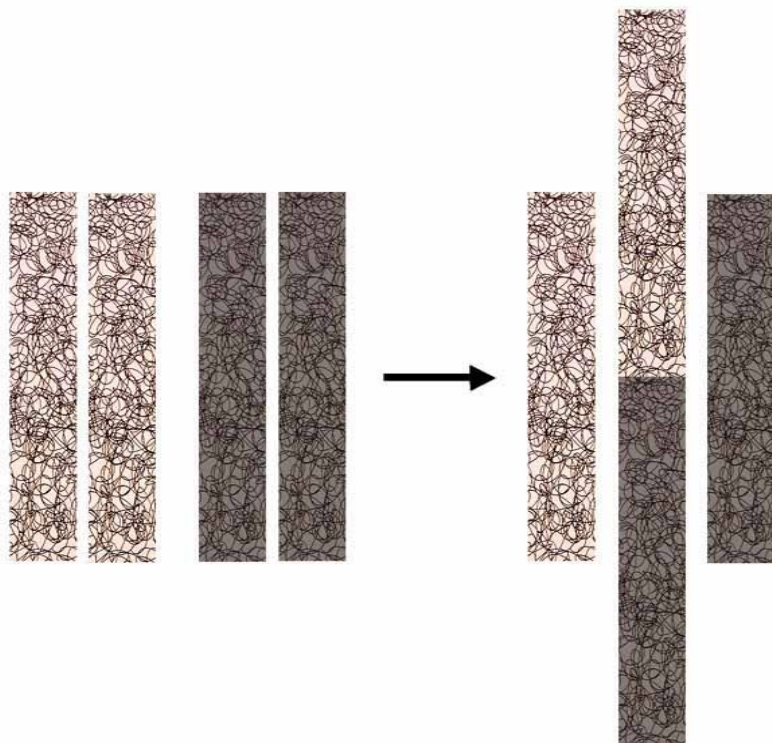
#### Picture 3: How a reciprocal translocation arises



## Robertsonian translocations

A Robertsonian translocation occurs when one chromosome becomes attached to another. Picture 4 shows a Robertsonian translocation involving two chromosomes.

### Picture 4: How a Robertsonian translocation arises



Two pairs of normal Chromosomes.....

Robertsonian translocation: a chromosome from one pair has become attached to a chromosome from another pair

## Why do translocations happen?

Although about 1 person in 500 has a translocation, we still do not really understand why they happen. We know that chromosomes seem to break and rejoin quite often during the making of sperm and eggs or around the time of conception, and it is only sometimes that this leads to problems. These changes occur without us being able to control them.

## When might this lead to problems?

In both the examples we have looked at, the chromosomes have been rearranged so that **no chromosome material has been lost or gained**. This is called a **balanced translocation**.

A person who carries a **balanced translocation** is not usually affected by it, and is often unaware of having it. The only time it may become important is when he or she comes to have children. This is because the child may inherit what we call an **unbalanced translocation**.

