WHAT ARE CHROMOSOME TRANSLOCATIONS?

This leaflet 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 child’s genetic specialist.

**ROBERTSONIAN TRANSLOCATIONS**

How a Robertsonian translocation arises

Two normal pairs of chromosomes

A chromosome from one pair has become attached to a chromosome from another pair
What are genes and chromosomes?

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

This picture shows a cell, DNA and chromosomes

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 inherit two copies of most genes, one copy from each parent. This is the reason we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

The chromosomes numbered one 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, the X chromosome and 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.

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 too much or too little genetic information can result in learning disabilities, developmental delay or health problems.

What is a translocation?

A translocation means that there is an unusual arrangement of the chromosomes. This can happen because:
— a change has arisen during the making of the egg or the sperm or around the time of conception.
— an altered chromosome arrangement has been inherited from either the mother or the father.

There are two main types of translocation: 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.

**Robertsonian translocations**

A Robertsonian translocation occurs when one chromosome becomes attached to another.

**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 if he or she has children. This is because the child may inherit what we call an unbalanced translocation.

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 disabilities, 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.

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 your genetic specialist.

What about other family members?
If a translocation is found, that person may wish to discuss this with other family members. This gives them 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
**RECIPROCAL TRANSLOCATIONS**

How a reciprocal translocation arises

Two normal pairs of chromosomes → Parts of two chromosomes break off → ... and attach to different chromosomes

**ROBERTSONIAN TRANSLOCATIONS**

How a Robertsonian translocation arises

Two normal pairs of chromosomes → A chromosome from one pair has become attached to a chromosome from another pair
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.

**Points to remember:**

— People who carry a balanced translocation are usually healthy. The only time that a problem may arise is if 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.

**Useful links**

[undiagnosed.org.uk/support-information](undiagnosed.org.uk/support-information)
[bit.ly/rrjoinswanuk](bit.ly/rrjoinswanuk)
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.

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