(2) It is important that children who carry, or may carry, a translocation know about it before they plan to have children of their own.

(3) People often feel guilty about something like a balanced translocation that runs in the family. It is important to remember that it is **no-one's fault**, and that no-one has done anything to cause it to happen. These things come "out of the blue" and are **out of anyone's control.**

(4) Sometimes, people find it difficult to tell other family members about translocations. Often they feel it is their duty to tell their relatives about the translocation, as they may be at risk of having a child with a serious disability, but they don't know quite how to go about this without causing undue worry. Some families are closer to each other than other families. Sometimes people may have lost touch with relatives and feel awkward about contacting them after so long. In such situations, it may be helpful to talk to someone from the genetic counselling clinic about how to approach family members, and which relatives in particular may need to know.

Therefore, if you, or any of your relatives, would like to talk about anything further, please feel free to get in touch with

---

at the address or telephone number below. We can always arrange an appointment.

For more information about Translocations:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)
Tel: 01224 552120 Fax: 01224 559390
(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)
Tel: 01382 632035 Fax: 01382 645731
(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)
Tel: 0131 651 1012 Fax: 0131 651 1013
(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)
Tel: 0141 201 0808 Fax: 0141 201 0361
(Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

This leaflet was written by Guy's & St. Thomas' Clinical Genetics. Updated by Genetic Interest Group Scotland.

Translocations

CLINICAL GENETICS CLINICAL GENETICS

Information booklet

Introduction

You may have been told that you, or a member of your family, has what is known as a **translocation** of the chromosomes. A translocation is an unusual arrangement of the chromosomes (or genetic material) in the cells of the body.

Most people have never heard of a translocation, and can feel worried if told that they have (or may have) something like this which is rather unusual. Understanding a little more about the translocation can often help to lessen such anxiety. We hope that this booklet will help to explain what this means, and answer some of your questions.

Genes and chromosomes: What are they and why are they important?

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

Genes are tiny 'packages' of vital information which influence our growth and development. Each person probably has about 30,000 genes, which all do something different. They work rather like special computer programmes, and determine such things as the colour of our eyes, how many fingers and toes we have, how tall and short we are, and so on. Genes are so small that they cannot be seen, even under a powerful microscope. They are arranged along little threadlike strands, rather like the way that beads are arranged along a string. These strands are called **chromosomes**, and are inside most of the cells in our body. Unlike genes, chromosomes can be seen under a microscope.

How many chromosomes does a person usually have?

Our bodies are made up of billions of cells. A person usually has a total of **46** chromosomes in each cell. They come in pairs. We inherit one of each pair of chromosomes from our mother and the other from our father. This is how we inherit characteristics from our parents.

The following picture shows what chromosomes would look like if we arranged them in pairs in order of their size. Each pair is numbered to help us identify them more easily.

Should other family members be told about the translocation?

If anyone in the family already has children, and/or is likely to have children in the future, it is usually sensible for them to be told. This gives them the opportunity to have a blood test to see if they also carry the translocation. If they do not carry the translocation, they cannot pass it on to any of their children, and the matter can be forgotten about from their point of view. If it is found that they do carry the translocation, they too could be offered a special test in pregnancy to check the baby's chromosomes, and any existing children can be offered genetic counselling and testing when they are older.

Children who carry (or may carry) a balanced translocation: why, and when, should they be told about this?

Why? Because a child who inherits the same balanced translocation as their carrier parent will have the same increased risk of having a disabled child when he or she comes to have a family of their own.

When? Each child is different, so there is really no one time when they 'should' be told that they do (or may) carry a translocation. It is probably sensible to leave this until they are old enough to be able to understand without being too worried by it. However, it may be best not to leave it until adolescence, when suddenly learning that you are 'different' from their friends can be very upsetting. Also, it is usually best for people to learn about the translocation well before having a family of their own. If any parent would like to talk more about this with us, we would be very happy to do so.

Can someone who 'carries' a translocation still be a blood donor?

Yes. A translocation is NOT a disease, and is not something that can be 'caught' from other people. It can only be passed from parent to child. Having a translocation should not stop you from giving blood if you want to do so.

And finally..... some important points to remember

(1) A balanced translocation of the chromosomes does **NOT** affect the health of someone who carries it. The only time it is important is when there is a pregnancy.

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 may have a disabled child is higher than average, although the precise risk depends on the exact type of the translocation.

Can a carrier of a balanced translocation have a special test in a pregnancy?

Yes. Some people who carry a balanced translocation choose to have a special test in pregnancy by which the chromosomes of a developing baby can be checked.

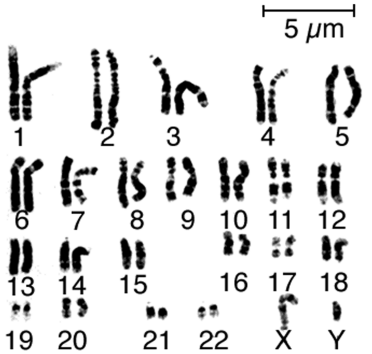
One of these is called an **AMNIOCENTESIS** test (there is a special leaflet available which explains this test in more detail). A sample of the amniotic fluid ("water") which surrounds the baby in the womb is withdrawn through a fine needle. This is usually performed from 16 weeks of pregnancy onwards. The result takes about 2-3 weeks to come through.

Another test is called **CHORIONIC VILLUS SAMPLING (CVS)** (there is a special leaflet available which explains this test in more detail). A sample of the developing placenta (afterbirth) is taken, either through the neck of the womb (rather like a cervical smear test) or through a needle in a similar way to an amniocentesis. The CVS test is usually performed from the 11th week of pregnancy until the 13th week of pregnancy, with the result taking 2-3 weeks.

If you would like more information about either of these tests, we would be pleased to send you some, or talk to you about the tests, preferably before a pregnancy. If, after one of these tests, the baby was found to have either the '**normal**' set of chromosomes or the same **balanced** translocation as the carrier parent, then nothing further would need to be done. However, if the baby was found to have an **unbalanced** translocation, the parents would then need to consider whether or not they wanted to continue with the pregnancy, or have a termination (abortion). This is because, sadly, there is no way of "correcting" the baby's chromosomes.

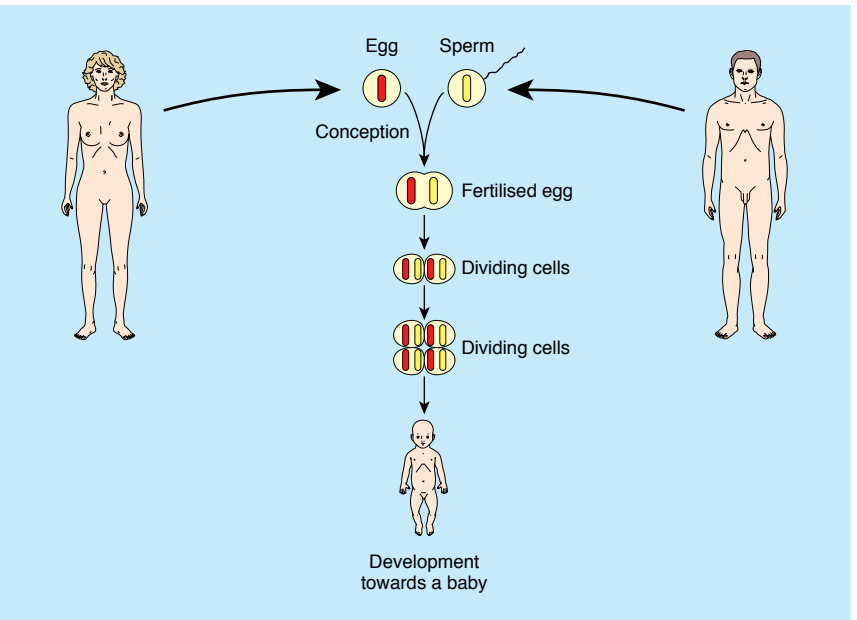
How can someone find out if they carry a balanced translocation?

A simple blood test is all that is needed. A small amount of blood is taken, and some of the blood cells examined in a special laboratory.



Pairs number "1" to "22" look the same in boys and girls. Pair number "23" are the sex chromosomes. They determine our sex. Girls have two "X" chromosomes, one inherited from their mother and one from their father. Boys inherit an "X" chromosome from their mother, and a "Y" chromosome from their father. The picture to the right shows chromosomes from a male.
At conception, the egg (with 23 chromosomes) joins with the sperm (also with 23 chromosomes) to make a fertilised egg. This divides and divides millions of times, and the baby develops.

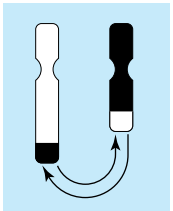
It is important that we have the correct amount of chromosome material, as the genes (which control the way we grow and develop right from when we are conceived) are found on the chromosomes. Having some part of a chromosome missing, or having an extra part of a chromosome, can therefore result in a problem with normal development and lead to physical disabilities and learning difficulties in a child.



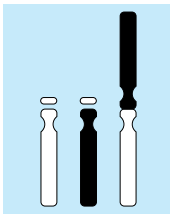
What happens in a translocation?

Sometimes, when cells are dividing during the formation of the egg or the sperm, or in the very early development of the baby, one or more of the chromosomes can break. This can cause a "translocation": an unusual arrangement of the chromosomes. There are two main types of translocation: a **RECIPROCAL** translocation and a **ROBERTSONIAN** translocation.

A **Reciprocal translocation** occurs when two fragments break off from two different chromosomes and "swap places", as shown in the diagram below.



A **Robertsonian translocation** occurs when two whole chromosomes become "stuck together". This diagram shows a Robertsonian translocation.



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, and it is only sometimes that this leads to problems. Translocations happen in either the egg or the sperm cell before they join together, or shortly afterwards. These changes are totally out of our control and are unlikely to be caused by anything that happens during a pregnancy.

To understand why translocations can be important, it is helpful to understand the difference between **BALANCED** and **UNBALANCED** translocations.

Balanced translocations

In both examples already shown in the pictures, the chromosome material has been rearranged in such a way that **no chromosome material has been lost or gained.** This is known as a **balanced translocation.**

The health of a person who "carries" such a balanced rearrangement of their chromosomes is **NOT** affected by it. The only time it is important to them is when they come to have children, when a baby can inherit what is called an unbalanced form of the translocation.

Unbalanced translocations

If one or other parent carries a balanced translocation, it is possible for a child of theirs to inherit a rearrangement of the chromosomes in which there is **an extra piece of one chromosome and/or a missing piece of another chromosome.** This is what is known as an **unbalanced translocation.**

As was explained earlier, having too little or too much chromosome material can result in disability. An **unbalanced** translocation can cause serious problems in the development of a child who inherits this kind of rearrangement. There are different sorts of disability, some more serious than others. The seriousness of the disability in individual cases may depend on exactly which chromosomes are involved, and how much of the chromosome material is missing or extra. Some parts of the chromosome do seem to be more important than others. However, if a baby does have an **unbalanced** translocation, there is nearly always some degree of disability.

If a parent has a balanced translocation, will he/she always pass it on?

Not necessarily. Each time a "carrier" of a balanced translocation has a pregnancy, there are several possibilities.

(1) A baby may have an entirely normal set of chromosomes.

(2) A baby may inherit the same balanced translocation as the parent. In this case the child would be expected to be a healthy carrier, and not be in any way disabled because of this.

(3) A baby may have an unbalanced translocation, and be mentally and physically disabled because of this.

(4) The pregnancy may end in a miscarriage.