Manchester University
NHS Foundation Trust

Saint Mary's Hospital
Manchester Centre for Genomic Medicine

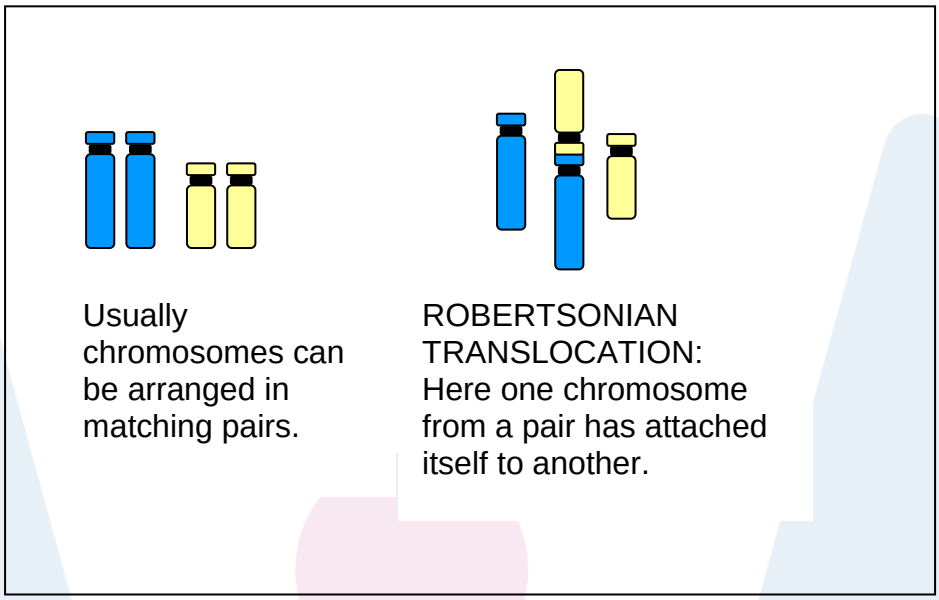
Information for Patients

Robertsonian Translocations

What happens in a translocation?

A robertsonian translocation is an unusual arrangement of the chromosomes. In around approximately 1 in 500 people when we look at the chromosomes, all the material is there but the pattern looks different.

When one chromosome from a pair has attached itself to the top of another chromosome this is known as a robertsonian translocation. Robertsonian translocations usually involve chromosomes from pairs 13, 14, 15, 21, or 22.



A person with a robertsonian translocation appears to have only 45 chromosomes when they are looked at under a microscope, but they still have the correct amount of chromosome material, it is simply that two chromosomes have joined together.

Why do translocations happen?

Approximately 1 in 900 people has a robertsonian translocation, but we do not really understand why they happen. Chromosomes break and re-join quite often, but it is only sometimes that they re-join in the wrong place to give a translocation.

Chromosome translocations occur in either the egg or the sperm cell. These changes are totally out of our control and are unlikely to be caused by anything that happens during a pregnancy. Once a translocation has occurred in a person it can be passed on to future generations. Some people carry a translocation which they have inherited from one of their parents. In others, it may have happened for the first time.

How can we find out if a person carries a translocation?

A simple blood test is all that is usually needed. The chromosomes inside the blood cells can be looked at down the microscope to see if they have the usual or a rearranged pattern. This test is offered to people who have a family history of a chromosome translocation or when there is suspicion of a chromosome rearrangement.

What does it mean to carry a translocation?

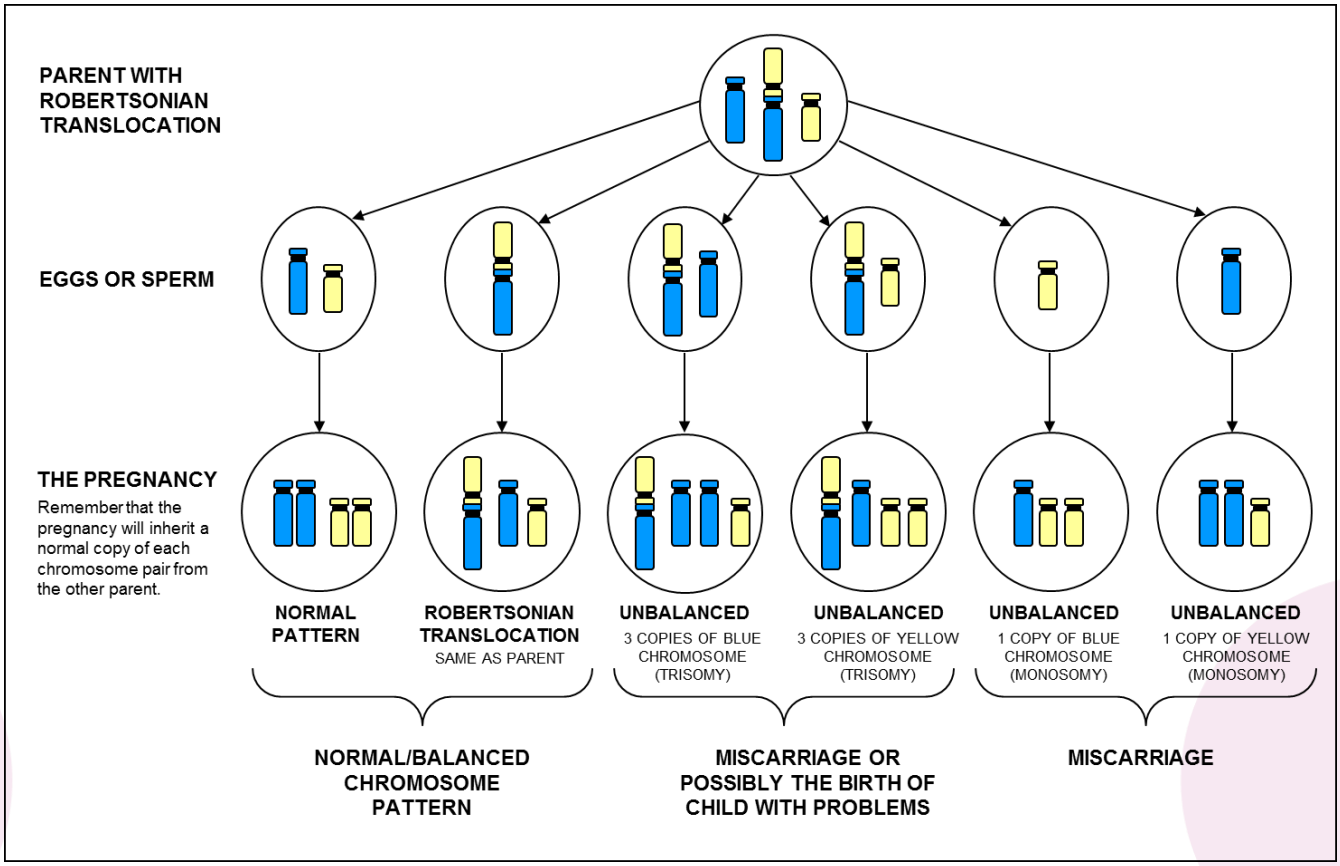
If there is no missing or extra chromosome material the translocation is balanced. People who have a balanced chromosome translocation have the right amount of genetic information even though this is arranged in a different way and there are no health implications for them.

A balanced translocation can cause problems when people come to have children, because there is a risk of passing on an unbalanced amount of genetic information. Having too little or too much chromosome material can result in disability. An **unbalanced** translocation may cause miscarriage or serious problems in the growth and development of the baby.

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 child with disabilities is higher than average. The possible outcomes in pregnancy for a robertsonian translocation carrier are described overleaf.

Possible chromosome patterns in a pregnancy

The diagram below shows some of the chromosome arrangements that can be passed on in pregnancy and the possible outcomes.



What is the effect of an unbalanced chromosome pattern?

In a pregnancy with an unbalanced form of the translocation, a copy of one of the chromosomes involved will be missing or extra. Having three copies of a chromosome instead of the usual two is called **trisomy**.

The effect of a trisomy depends on which chromosome is involved. If it is chromosome 14, 15 or 22 it is likely that the pregnancy would miscarry at an early stage. If it is 13 or 21, the pregnancy may miscarry or continue to term, resulting in the birth of a child with problems. Trisomy 21 is also known as Down syndrome. Trisomy 13 is called Patau syndrome.

Having one copy of a chromosome instead of the usual two is called **monosomy**. In this case there is not enough genetic material and the pregnancy would miscarry at an early stage.

Can we tell which chromosome pattern will be passed on?

Unfortunately, it is impossible to predict how often a certain pattern will be passed on and often we are not able to detect any problems with a baby on early pregnancy scans.

It is possible to have invasive prenatal testing in pregnancy to detect the chromosome pattern. There are two types of test: chorionic villus sampling (CVS) and amniocentesis. There is a small risk of miscarriage after both tests. Separate information leaflets are available about both CVS and amniocentesis. The choice to have these tests is individual and the exact risks of your specific translocation should be discussed in your genetic counselling appointment.

If an unbalanced chromosome pattern is found, this could lead to a miscarriage or to the birth of a baby with serious disability. In these circumstances, a couple may consider whether to continue with the pregnancy. If, however, the pregnancy was found to have the 'usual' or the 'balanced' chromosome pattern, we would not expect there to be any increased risk of problems for the baby.

Are there any other reproductive options?

In some families where the risk of having a child born with significant problems is high a process called Pre-implantation Genetic Diagnosis (PGD) can be an option. This is an assisted reproductive technique and has some similarities to IVF but involves testing of the embryos before they are transferred to the womb. The aim of this process is to avoid using any embryos that have an unbalanced chromosome translocation. While this can sound appealing, it is not a straightforward technique and needs to be considered carefully for each couple. If this is something you are interested in it can be discussed in more detail with your genetic health professional. A separate leaflet is available.

People with genetic conditions in the family can also choose to have no children, adopt or have natural pregnancies with no testing involved.

When should we talk about the translocation with children?

There is no one time when children should be told about the family translocation since all children are different.

Children who carry a balanced translocation have no increased risk of health problems themselves. However, when they grow up, they will have an increased risk of experiencing problems in their own pregnancies. It is usually best for young people to learn about the translocation before they are considering 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.

Should other family members be told about the translocation?

If anyone in the family has children, or is likely to have children, then it is important that they are told about the translocation in the family. This gives them the opportunity to consider having a blood test to find out if they carry the translocation.

Sometimes, for many reasons, people find it difficult to tell family members about the translocation. If you would like to talk to someone about the best way to approach family members and which relatives may need to know, our Genetic Counsellors have a lot of experience with families in these situations.

Some important points to remember:

- We would not expect a robertsonian translocation to affect the health of anyone who carries it. The only time it is important is when there is a pregnancy.
- A carrier of a balanced translocation **can** have healthy children.
- It is important that other family members are told about the translocation. Children who could carry the translocation should be told about it before they plan to have children of their own.
- People often feel guilty about something like a robertsonian 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.

For more information

Please refer to our separate leaflet '*An Introduction to Chromosomes'*. If you need more advice about any aspect of robertsonian translocations, you are welcome to contact:

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Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally designed and wrote this leaflet.