

# Robertsonian Translocations

## Information Guide

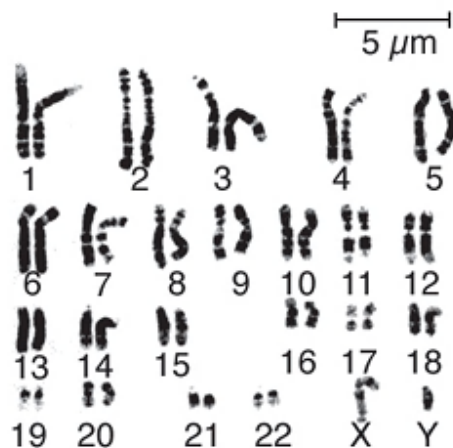
### Introduction

You, or a member of your family, may have been told that you have a Robertsonian chromosome translocation. This leaflet aims to answer some of the questions that you might have about what the Robertsonian translocation means for you and your family.

### What are genes and chromosomes?

Chromosomes are packages of genes. Genes are instructions which determine everything about us, from the colour of our hair to how tall we are. There are hundreds of genes along each chromosome.

Every cell in our body contains a set of 46 chromosomes. These chromosomes each have a matching partner. This picture shows the chromosomes from one cell arranged and numbered in pairs according to their size.

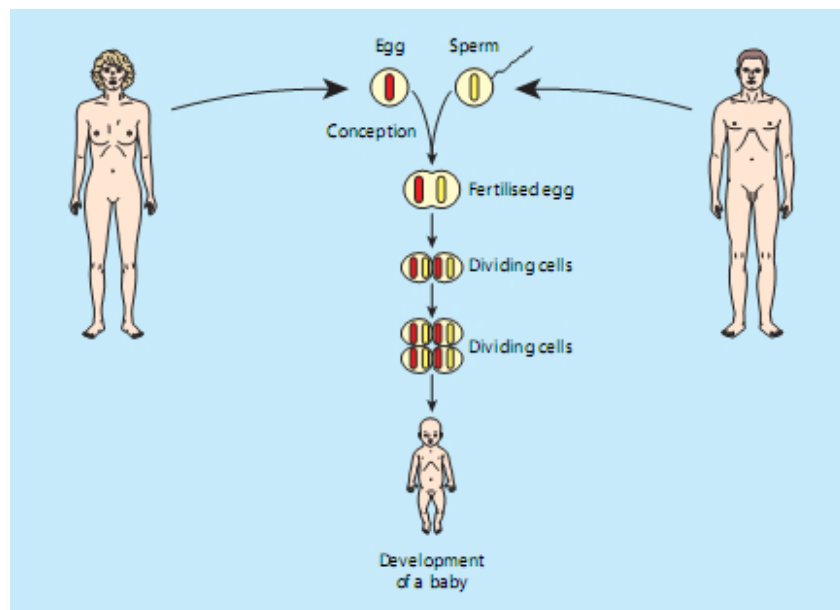


This male has one X and one Y chromosome. A female has two X chromosomes and does not have a Y chromosome. The remaining 22 pairs of chromosomes are the same in males and females.

## How do we inherit genetic information?

We inherit one chromosome of each pair from our mother and the other from our father.

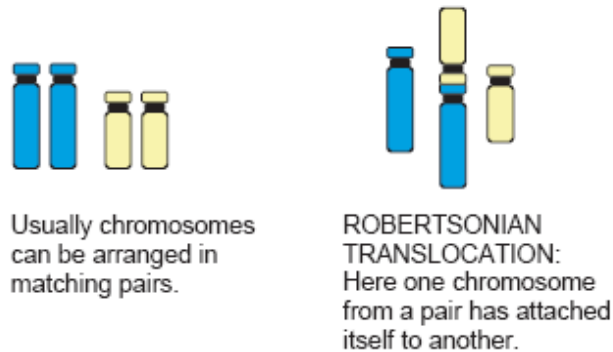
When we have children we pass on one chromosome of each pair in the sperm or egg. The sperm and egg each contain 23 chromosomes. After the sperm and the egg have joined, the fertilized egg contains 46 chromosomes in 23 matching pairs. This is shown in the picture below.



It is important that we have the right amount of genetic material as the genes control the way we grow and develop. Having too much or too little genetic material usually causes significant problems in development.

## What happens in a translocation?

A chromosome translocation is an unusual arrangement of the chromosomes. It was first described by a Dr Robertson who gave the chromosome rearrangement his name.



Robertsonian translocations usually involve chromosomes from pairs 13, 14, 15, 21 and 22. A person with a Robertsonian translocation appears to have only 45 chromosomes when the scientists look at their cells under a microscope. In fact, they have the correct amount of chromosome material, it is simply that two chromosomes have joined together.

## Why do translocations happen?

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

Chromosome translocations occur in either the egg or the sperm cell before fertilization or shortly afterwards. 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.

## What does it mean to carry a translocation?

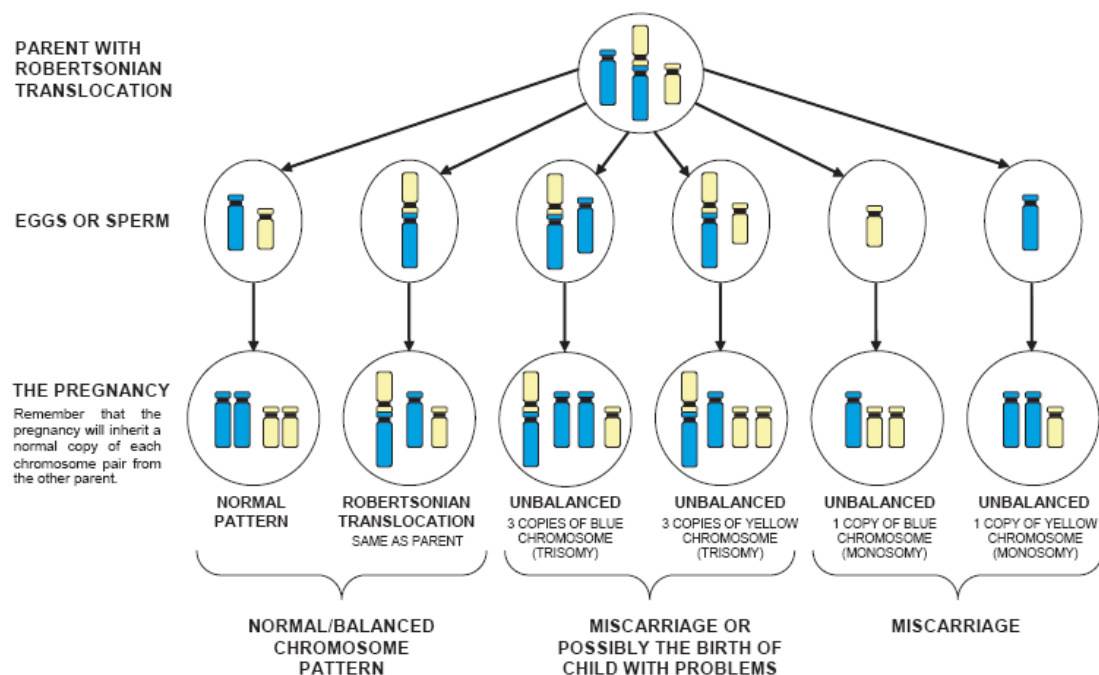
If there is no missing or extra chromosome material the translocation is **balanced**. People who have a balanced chromosome translocation have the right amount of genetic information even though this is arranged in a different way. They are as healthy as anybody else.

However, someone with a balanced translocation can experience problems when they come to have children. This is explained in more detail later in this leaflet.

## Can a person who carries a translocation pass it on to their children?

Yes. Translocations can be passed on. There are a number of ways that translocation carriers can pass on their chromosomes. This is shown below.

If one or other parent carries a Robertsonian translocation, a child may inherit a normal or a balanced Robertsonian pattern. However, it is also possible that they could inherit an **unbalanced** pattern. This means there is too much or too little chromosome material. Having too little or too much chromosome material can result in disability. An unbalanced translocation may cause miscarriage or problems in the growth and development of the baby.



## 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 we realise this can be difficult. It is quite possible for a person who carries a balanced Robertsonian translocation to have healthy children and many do. The chance of a pregnancy inheriting an unbalanced form of the translocation will depend on the chromosomes involved and whether the mother or the father is the translocation carrier. We will be able to discuss your particular situation with you in more detail in the genetics clinic.

### **Can a carrier of a balanced translocation have testing in pregnancy?**

Yes. Some people who carry a balanced translocation choose to have a test in pregnancy to determine the chromosome pattern of the developing baby. There are two procedures that can be used to obtain a sample. There are called chorionic villus sampling (CVS) and amniocentesis. The two procedures are performed at slightly different stages of pregnancy and each has its own advantages and disadvantages. There is a small risk of miscarriage after both tests.

If you would like to know more about these tests in pregnancy, we can discuss them at your clinic appointment. Separate information leaflets are available about both CVS and amniocentesis.

### **What happens if the chromosomes in the pregnancy are unbalanced?**

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 the couple may consider whether to continue with the pregnancy. Sadly, there is no way of correcting the chromosomes in the pregnancy.

If, however, the pregnancy was found to have the 'normal' or the 'balanced' chromosome pattern, we would not expect there to be any increased risk of problems for the baby.

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

A simple blood test is all that is usually needed. The chromosome inside the blood cells can be looked at with a microscope to see if they have a normal or rearranged pattern.

### **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. If they are found not to carry the translocation, then they cannot pass it on to their children. However, if they are found to be a carrier, they too could be offered testing in pregnancy to check the baby's chromosomes and any existing children can be offered genetic counselling and testing when they are older.

### **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 Robertsonian 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 probably sensible to wait until children are old enough to be able to understand the information without being too worried by it. However, it is usually best for young 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.

### **What service can we offer families with a translocation?**

The Genetic Medicine department at St Mary's Hospital has a Genetic Family Register Service for families with a translocation.

If you are found to carry a translocation, you and your family will be asked if you wish to be included in our Register Service. If you do, we will stay in contact with you for as long as you wish to remain on the Register.

We will send you a letter every few years to offer you an appointment to discuss any concerns you may have. If you have any questions or you become pregnant between these times you can always phone the department to arrange an appointment. We can also offer genetic counselling, where appropriate, to other family members.

The Register can also provide an opportunity to take part in any research that may help us to improve our understanding of translocations. You are not obliged to take part in any research and your treatment will not be affected if you refuse.

### **Some important points to remember**

1. We would not expect a balanced Robertsonian translocation to affect the health of anyone who carries it. The only time it is important is when there is a pregnancy.
2. A carrier of a balanced Robertsonian translocation **can** have healthy children.

3. 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.
4. 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.
5. Sometimes people find it difficult to tell family members about the translocation. They know their relatives may be at risk of having a child with a serious disability. They may feel it is their duty to talk to their relatives about the translocation, but they don't know how to go about this without causing undue worry. In some families, people have lost touch and may feel awkward about getting in contact again. 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.

### For additional information or support

If you have any further questions about Robertsonian translocations, please contact:

#### Genetic Medicine

6<sup>th</sup> Floor, St Mary's Hospital, Oxford Road, Manchester M13 9WL

Telephone 0161 276 6506

Fax 0161 276 6145

Department staffed Monday–Friday, 9.00am to 5.00pm.

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আপনি কি ইংরেজীতে বুঝতে কিংবা বুঝাতে পেরেছেন ? (BENGALI)

क्या आपको अंग्रेजी बोलने या समझने में कठिनाई है ? (HINDI)

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ਕਿ ਤੁਹਾਨੂੰ ਅੰਗ੍ਰੇਜ਼ੀ ਬੋਲਣ ਜਾਂ ਸਮਝਣ ਵਿਚ ਦਿੱਕਤ ਹੈ ? (PUNJABI)

Miyey ku adagtahay inaad ku hadasho Ingriisida aad sahamto (SOMALI)

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