

# Robertsonian Translocations

An information leaflet for  
parents and families

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## Introduction

This leaflet has been produced for families where a condition called a 'balanced translocation' has been found. This leaflet describes what happens when someone has a **Robertsonian** translocation. We hope that you find it useful and will answer some of your questions.

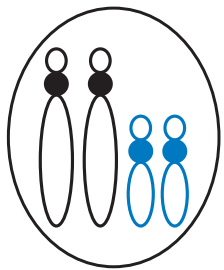
## What is a chromosome?

We have a complete set of chromosomes in every cell in our body and they contain the information the body needs to develop and function properly. This information is carried in the genes which are arranged within the chromosomes.

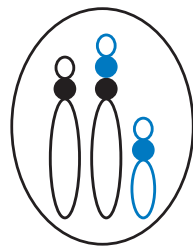
A person usually has 46 chromosomes in each cell. Chromosomes come in pairs and we inherit one of each pair from our father and one of each pair from our mother.

## What is a Robertsonian translocation?

A translocation is an unusual arrangement of the chromosomes in the cell. Most people with a translocation have the correct amount of genetic material but it is arranged in an unusual pattern. In the case of Robertsonian translocations, two whole chromosomes become 'stuck together' resulting in 45 chromosomes instead of the usual 46.



Usual chromosome  
pattern



Robertsonian chromosome  
pattern

## Further information

Clinical Genetics Unit

<http://bwhct.nhs.uk/genetics-wmrcgs-home.htm>

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## Balanced Translocations

A balanced translocation is where all the genetic information is present but not quite in the usual pattern. This has no effect on health but can cause reproductive problems such as recurrent miscarriages and fertility problems.

## Unbalanced Translocations

An unbalanced translocation is where there is too much or too little genetic material. This can lead to a miscarriage or even the birth of a child with physical and learning difficulties.

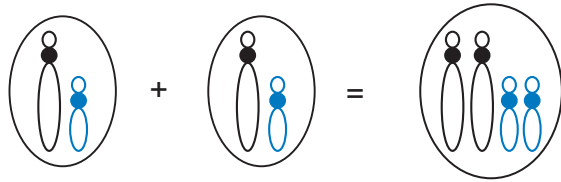
## I'm a Robertsonian Translocation carrier – what does that mean for me?

Translocations are common. It is thought that about 1 in a 1000 people have a Robertsonian translocation. Providing there is no loss or gain of chromosome material carrying a translocation will not affect your health. People with balanced translocations may have reproductive problems.

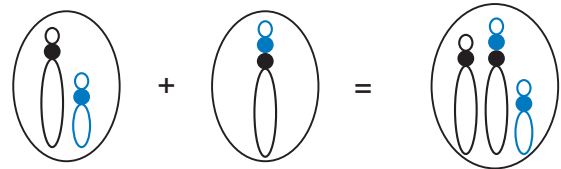
## What happens when a Robertsonian Translocation carrier has children?

One parent will pass on a usual set of chromosomes and the parent with a translocation can pass on a combination of chromosomes. There are a number of possibilities that can happen. The child may have normal chromosomes as shown on the next page.

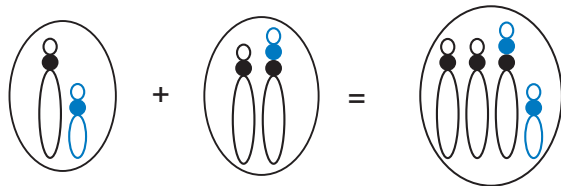
Usual chromosome pattern



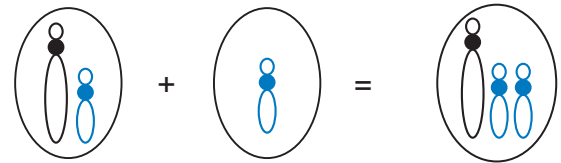
The child may have a balanced translocation - the same as the parent.



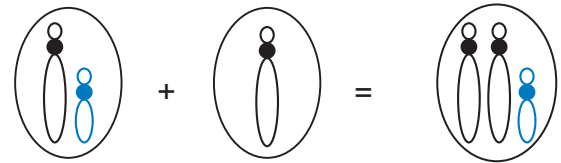
An unbalanced form may be inherited, shown below, whereby there may be too much or too little genetic material.



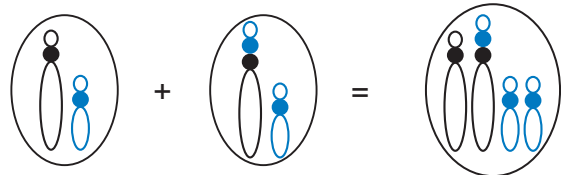
or



or



or



An unbalanced translocation may result in a miscarriage or even the birth of a child with developmental and physical problems.

## What tests are available in a pregnancy?

It is possible to have a test in pregnancy to check the baby's chromosomes. There are two tests available. These are called a CVS test (chorionic villus sampling) and an amniocentesis.

Your genetic counsellor can give you more information about these tests if you wish.

## How do I find out if I have a Robertsonian translocation?

A simple blood test can identify whether or not you are a carrier of a Robertsonian translocation.

This can be arranged via your local Genetics service. Results can take around 6 weeks.

## Should I tell other family members?

When someone is found to have a balanced translocation the most likely explanation is that it has come from one of the parents. Because translocations can be inherited it is important that other members of the family are given the opportunity to be tested.

Occasionally a translocation occurs which has not been inherited from either parent. This is a new rearrangement that has occurred in the egg or sperm cell that made that person. This is known as a de novo translocation.

## When should I tell my children?

If you are found to be a carrier then there is also a chance that your children are translocation carriers. As being a carrier does not have any affect on your health, we advise that the children are given the opportunity to discuss their risk and have the option of being tested around the age of 16 years.