

Robertsonian Translocations

Information for patients

What are chromosomes?

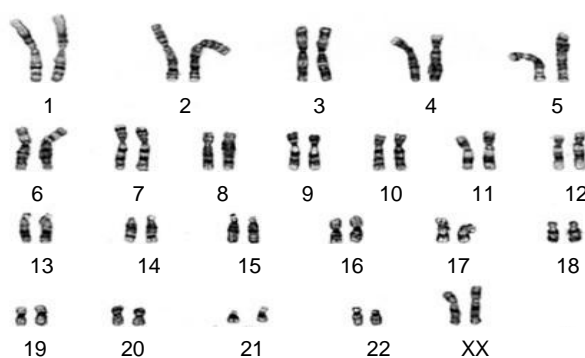
We are all made up of tiny building blocks called cells. These cells are controlled by information stored in long thin strands of DNA (DeoxyriboNucleic Acid) which are stored in the nucleus (control centre) of the cell. The DNA controls the cells using signals from about 30,000 genes. Each gene is a specific piece of DNA that contains particular genetic instructions for our body to grow and develop.

The DNA is normally seen in 46 strands called chromosomes which are arranged into 23 pairs. Scientists have numbered these pairs from 1-22 according to their size, with chromosome 1 being the longest and chromosome 22 the shortest.

The two chromosomes in each of these pairs are usually identical. The 23rd pair is called the sex chromosomes which determine whether we are male or female. Females have two X chromosomes and males have an X and a Y chromosome.

Each chromosome has a short arm and a long arm, separated by a narrow area in the middle. Scientists only need a small amount of blood to look at a person's chromosomes under the microscope. The picture below shows what our chromosomes look like under the microscope if they are arranged according to their size.

Chromosomes from a normal female:



How are chromosomes inherited?

When eggs or sperm are made, the pairs of chromosomes separate and one from each pair goes into each egg or sperm. Therefore, eggs and sperm only have 23 chromosomes.

All eggs have an X sex chromosome and each sperm has either an X or a Y sex chromosome. When the egg is fertilised by the sperm, the resulting baby inherits half of its chromosomes, and therefore its genetic material, from its mother and half from its father.

We need to have the correct amount of genetic material for normal development. At any stage during this process, mistakes can happen. These can involve:

- The wrong number of chromosomes (either extra or missing chromosomes). A common example is Down's syndrome, where individuals have three copies of chromosome 21.
- Mistakes in the structure of the chromosomes such as translocations.

What is a Robertsonian translocation?

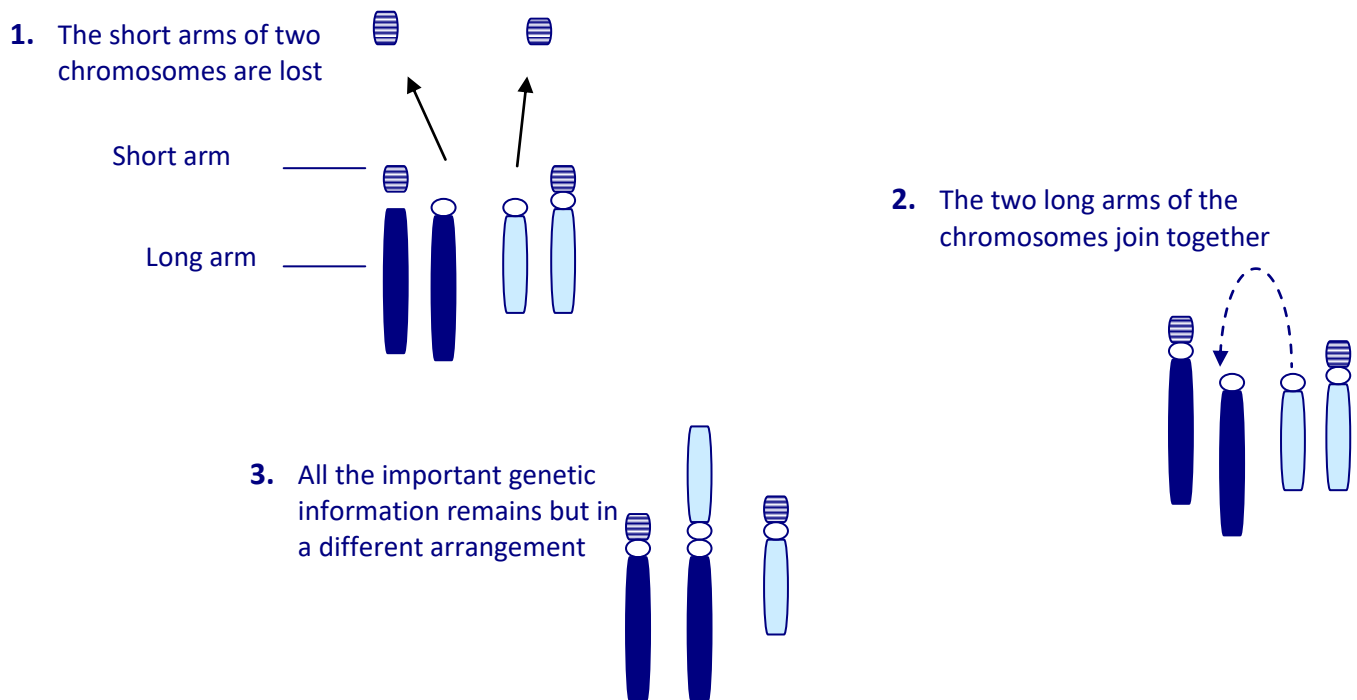
A translocation means that a piece of one chromosome has broken off and stuck on a different chromosome. Chromosome translocations cannot be repaired. If a translocation has not resulted in any genetic material being lost, it is known as a balanced translocation. Balanced translocations do not usually cause any medical problems. This is because all the genetic material is present, even though it is in a slightly different arrangement.

Robertsonian translocations are a particular type of balanced translocation that get their name from an American scientist who first described them.

Robertsonian translocations only involve certain chromosomes (numbers 13, 14, 15, 21, 22). These chromosomes are different from the other chromosomes because they have a long arm and a very short arm.

In a Robertsonian translocation, the short arms of two of these chromosomes are lost and the remaining long arms join together. As the short arms of these chromosomes do not contain important genetic information, this translocation is described as balanced and has no effect on a person's health.

The picture below shows how a Robertsonian translocation happens.



Robertsonian translocations occur in about 1 in 1000 people.

The most common Robertsonian translocation is between chromosomes 13 and 14, accounting for $\frac{3}{4}$ (75%) of all these translocations.

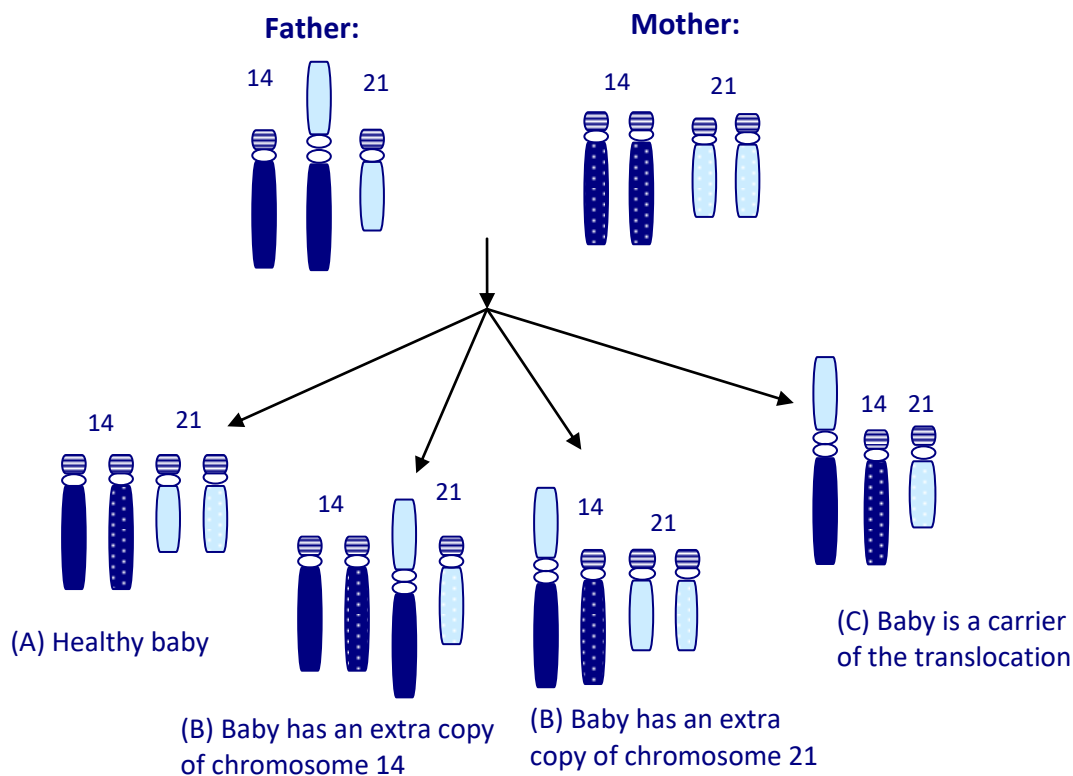
Having children

The main concern for people with a Robertsonian translocation is that they may have a child with extra genetic material, which can cause medical problems. For each pregnancy, the outcome depends on whether the sperm or the egg from the parent who has the Robertsonian translocation contains the Robertsonian translocation and/or normal chromosomes.

For example, if the father has a Robertsonian translocation involving chromosomes 14 and 21 and the mother has normal chromosomes, there are different possibilities as shown in the picture on the next page. However, it is not possible to determine the likelihood of each of these possibilities.

The baby can inherit:

- A. Neither of the chromosomes from the father that were involved in the Robertsonian translocation. This will result in a normal healthy baby as they have inherited a normal set of chromosomes
- B. The father's translocated chromosomes and one of the father's other normal chromosomes and normal copies of the chromosomes from the mother. This means that the baby has extra genetic material (i.e. the translocation is now unbalanced). This is likely to result in physical or mental disability. The type and severity of the disability depends on the extra chromosome. If the baby has an extra copy of chromosome 14, making 3 copies instead of 2 (this is known as Trisomy 14), the pregnancy is likely to end in an early miscarriage. If the baby has 3 copies of chromosome 21 (Trisomy 21), they will have Down's syndrome.
- C. The father's translocated chromosomes and normal copies of the chromosomes from the mother. This does not usually cause any medical problems, but the baby will be a carrier of the Robertsonian translocation just like their father.



What are the implications?

A person with a Robertsonian translocation has an increased risk of miscarriage for each pregnancy and an increased risk of the baby being born with a disability.

There are special tests in pregnancy that can check the baby's chromosomes. Carriers of a Robertsonian translocation can choose to have these tests to help them make decisions about the pregnancy. A local genetic clinic can discuss options for planning a family in more detail.

Some men with a Robertsonian translocation experience fertility problems caused by a low sperm count or problems with sperm production. However, this does not happen in all men with a Robertsonian translocation.

Often it is only when a baby is born with extra genetic material that one of the parents is found to have a Robertsonian translocation. As there is a possibility that the Robertsonian

translocation may run in their family, other family members may benefit from knowing whether they have the Robertsonian translocation, particularly if they are likely to have children in the future. In this instance, they should be referred to their local clinical genetics service for advice.

Where can I find more information?

Unique is a source of information, mutual support, and self-help to families of children with any rare chromosome disorders including translocations.

Telephone Helpline: 01883 330766

Website: www.rarechromo.org/

Your local genetics service:

South East of Scotland Clinical Genetic Services
The Institute of Cancer and Genetics
Western General Hospital
Crewe Road South
Edinburgh
EH4 2XU

Telephone: 0131 537 1116