

# RECIPROCAL TRANSLOCATIONS

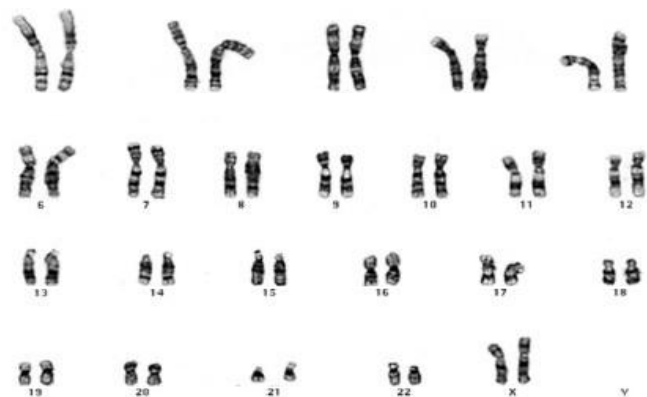
## INFORMATION FOR PATIENTS

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 23<sup>rd</sup> pair are 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 opposite shows what our chromosomes look like under the microscope if they are arranged according to their size.

Chromosomes from a normal female:



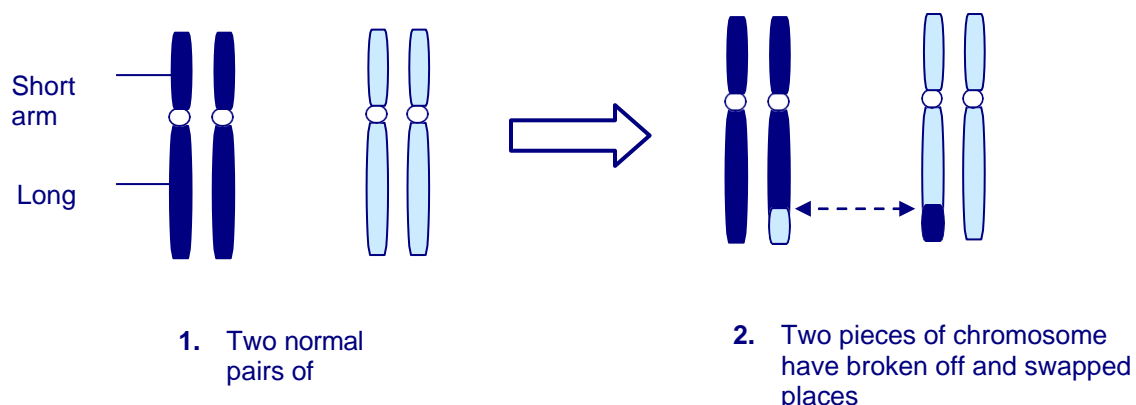
## HOW ARE CHROMOSOMES INHERITED?

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.

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. The picture below shows how a balanced translocation happens.



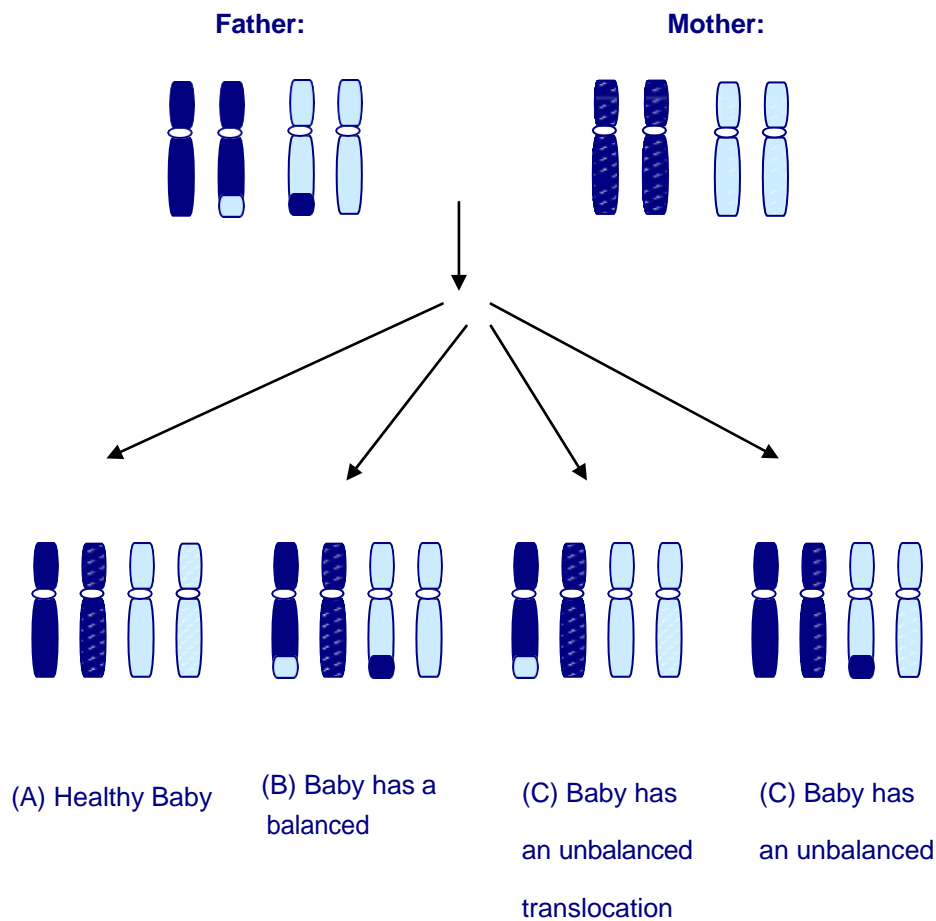
Balanced translocations do not usually cause any medical problems. This is because all the genetic material is present, even though it is in a slightly different arrangement. Balanced translocations are relatively common and occur in 1 in 625 people.

The main concern for people with a balanced translocation is that they may have a child with missing or 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 balanced translocation contains both, one or none of the chromosomes that was involved in that translocation.

For example, if the father has a balanced translocation between chromosomes 3 and 7 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 balanced translocation. This will result in a normal healthy baby as they have inherited a normal set of chromosomes.
- B. Both chromosomes from the father that were involved in the balanced translocation. This does not usually cause any medical problems, but the baby will be a carrier of the balanced translocation just like their father.
- C. One chromosome from the father that was involved in the balanced translocation and normal copies of the other chromosomes. This means that the baby has missing or 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 amount of genetic material that is missing or extra. If a large amount of genetic material is missing or extra, the pregnancy is likely to end in a miscarriage.



For each pregnancy, a person with a balanced translocation has an increased risk of miscarriage 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 balanced translocation can choose to have these tests to help them make decisions about the pregnancy.

Often it is only when a baby is born with missing or extra genetic material that one of the parents is found to have a balanced translocation. As there is a possibility that the balanced translocation may run in their family, other family members may benefit from knowing whether they have the balanced translocation, particularly if they are likely to have children in their 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: +44 (0)1883 723356

WEBSITE: [HTTP://WWW.RARECHROMO.ORG/](http://www.rarechromo.org/)

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### Your local genetics service:

South East of Scotland Clinical Genetic Service:	MMC, Western General Hospital Crewe Road South, Edinburgh EH4 2XU Telephone: 0131 537 1116
North of Scotland Genetics Service:	Department of Clinical Genetics Ashgrove House, Foresterhill Aberdeen AB25 2AZ Telephone: 01224 552120
East of Scotland Genetics Service:	Human Genetics Unit Level 6, Ninewells Hospital Dundee DD1 9SY Telephone: 01382 632035
West of Scotland Genetics Service:	Level 2A Laboratory Medicine The Queen Elizabeth University Hospital 1345 Govan Road, Glasgow G51 4TF Telephone: 0141 354 9201