Robertsonian Translocation

What are genes and chromosomes?

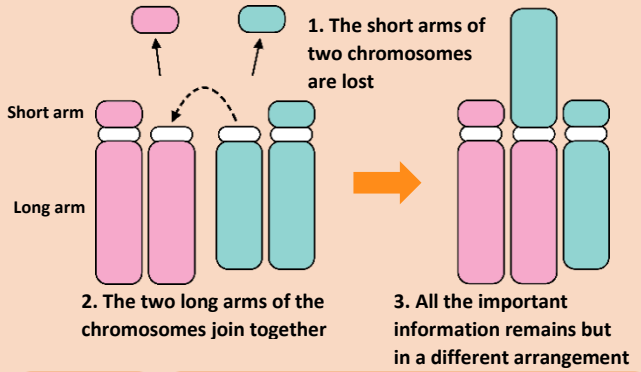
The human body is made up of billions of cells. A person usually has 46 chromosomes in each cell (arranged in 23 pairs). A pair of chromosomes that are basically identical in structure are called homologous chromosomes. One of each pair of chromosomes is inherited from the mother, and one from the father. Most chromosomes carry hundreds to thousands of genes that provide instructions to determine an individual's physical traits and help the body to grow and function properly. It is important to have the correct amount of chromosomal material for normal development.

What is a Robertsonian translocation?

Chromosome translocation is an unusual arrangement of the chromosomes in the cell. Robertsonian translocation is an unusual type of chromosome rearrangement caused by the fusion of two acrocentric chromosomes, the rearrangement commonly involves chromosomes 13, 14, 15, 21, and 22. About 1 in 1000 people has a Robertsonian translocation and rearrangement between chromosomes 13 and 14 accounts for 75% of all Robertsonian translocations.

In a Robertsonian translocation, the short arms of two affected chromosomes are lost and the remaining long arms join together. As the short arms of the chromosomes do not contain important genetic information, this translocation is described as **balanced Robertsonian translocation**.

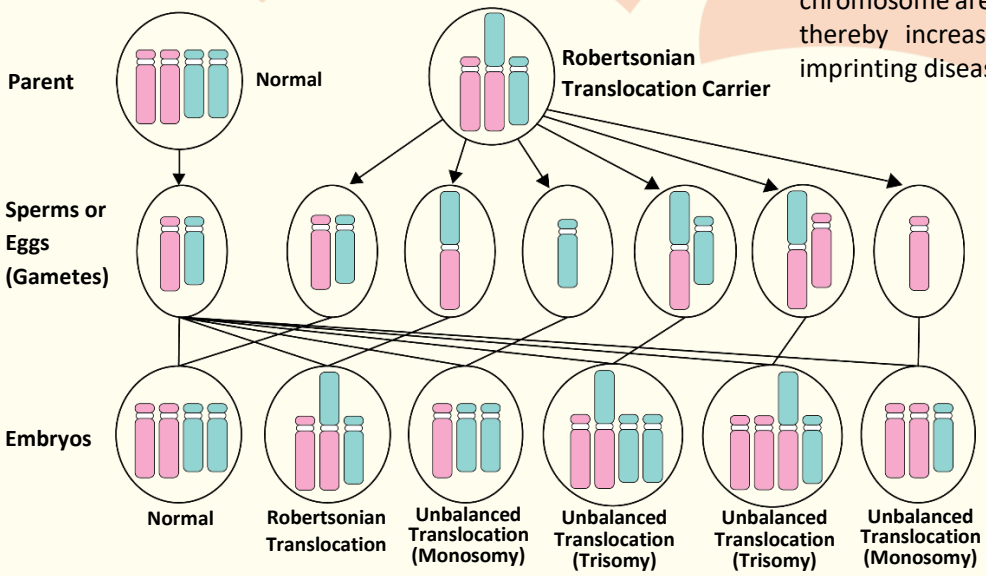
The picture below shows how a balanced Robertsonian translocation happens:



Does a Robertsonian translocation affect health?

People who carry the chromosome with balanced Robertsonian translocation (a carrier) usually have no health or developmental problems, but it may become important when they come to have children as there is a possibility a fetus/baby can inherit an unbalanced translocation with missing or extra genetic materials. An unbalanced translocation may end in miscarriage or the birth of a child with congenital abnormalities and/or learning difficulties.

Gene Map of Robertsonian Translocation



Can a balanced Robertsonian translocation carrier have children?

Although the carrier of balanced Robertsonian translocation has an increased risk for infertility, recurrent miscarriages, or having children with congenital abnormalities, most men and women with a balanced Robertsonian translocation can have children. There are usually five possible outcomes if one parent is a carrier of balanced Robertsonian translocation:

(1) A baby inherits a normal set of chromosomes.
(2) A baby inherits the same balanced Robertsonian translocation as the parent. The baby would be expected to be a healthy carrier, similar to their parent.
(3) A baby inherits the unbalanced translocation with either too much or too little genetic material which results in physical and/or learning problems.
(4) A fetus inherits the unbalanced translocation with either too much or too little genetic material which disrupts the fetus's development. The pregnancy may end in a miscarriage.
(5) Increased risk of a fetus suffering from uniparental disomy (UPD). Both copies of chromosome are inherited from only one parent, thereby increasing the chance of developing imprinting disease.

❖ *Translocation can occur in familial or sporadic form. For sporadic form, it is occurred in the egg or sperm cell that made that person which is called a de novo translocation.*

How to detect chromosome translocations?

Chromosome translocations can be detected by a genetic test called karyotyping. It is a simple test that examines the chromosomes inside the cells from the blood or other specimens (like amniotic fluid) to look at any unusual chromosomal arrangement. The test can be performed to detect the karyotype of an individual or fetus.

Balanced Robertsonian translocation carriers do not have any physical signs or symptoms, so most of the carrier are unaware of their conditions until parental karyotyping or karyotyping of either aborted fetus or affected newborn is performed. Because balanced Robertsonian translocation carriers are at higher risk of recurrent miscarriage, therefore, karyotyping is recommended for couples who have had three or more pregnancy losses to detect whether they are carriers of balanced Robertsonian translocation.

What can be done if you have a balanced Robertsonian translocation?

Although a balanced Robertsonian translocation cannot be corrected and does **NOT** affect the health of the carriers, genetic counselling is recommended for the carriers when planning for a pregnancy. The results of genetic testing may help to clarify the risks and the options for the next pregnancy.

Depending on the genetic test results, the couples may consider:

- Prenatal diagnosis for the fetal chromosomal analysis through chorionic villus sampling or amniocentesis when the couples conceive again.
- *In vitro fertilization* with special genetic testing called preimplantation genetic diagnosis (PGD) may be done to select unaffected embryos.

Should I tell other family members if I am a carrier?

As Robertsonian translocation is a genetic condition, other family members may have inherited the same conditions. They may benefit from knowing whether they have a balanced Robertsonian translocation, particularly if they are likely to have children in the future. In this instance, it is recommended for them to seek medical advice from healthcare professionals.

For further enquires, you may contact us.

Department of Clinical Genetics

Specialist Outpatient Clinic,
1/F, Tower B,
Hong Kong Children's Hospital
1 Shing Cheong Road, Kowloon Bay,
Kowloon, Hong Kong
Tel: 5741 3186

香港兒童醫院
Hong Kong Children's Hospital

GENETICS AND GENOMICS CLINIC

DEPARTMENT OF CLINICAL GENETICS

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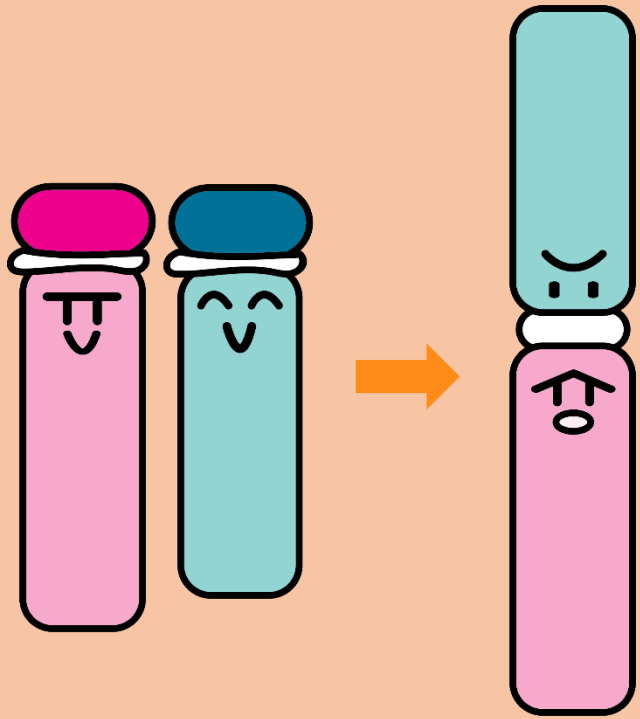

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