

What happens during pregnancy?

Some people choose to have testing during pregnancy to find out whether the baby has inherited the unbalanced form of the translocation.

This can be done by amniocentesis or Chorionic Villus Sampling (CVS). Please see Unique's 'Balanced Translocations' information guide for more information about this (which can be downloaded from the website: www.rarechromo.org).

Why me? Is it my fault?

If you've been told you have a balanced insertional translocation, you may wonder 'Why me?' Remember that the translocation probably arose when the egg or sperm cell that was destined to create you was formed. At the very latest, it arose in the earliest days of your mother's pregnancy. So there is certainly nothing you can blame yourself for or feel guilty about. Everything that is known about balanced insertional translocations suggests that it is chance whether you have one or not. No environmental, diet, workplace or lifestyle factors are known to cause them. They affect men and women from all types of background and all income and ethnic groups. They are not your fault and they are not the fault of anyone else in your family. Some people with a balanced insertional translocation feel guilty. Some people who inherit a translocation find that the parent who passed it on feels guilty. There is absolutely no reason to do so.

Unique members with balanced insertional translocations

At June 2014, Unique has around 40 members with a balanced insertional translocation, involving various different chromosomes. Children who have inherited an unbalanced form of the translocation usually have physical and/or learning problems. For more information regarding a particular duplication or deletion caused by inheritance of an unbalanced insertional translocation, please see our information guides (which can be downloaded for free from www.rarechromo.org) or contact Unique.

Inform Network Support

Join Unique for family links, information and support

Unique is a charity without government funding, existing entirely on donations and grants. If you can please make a donation via our website at www.rarechromo.org
Please help us to help you!



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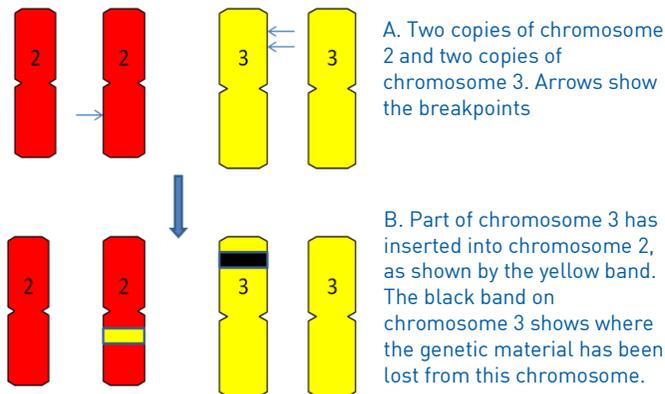


Balanced Insertional Translocations

What is a balanced insertional translocation?

Our bodies are made of billions of cells. In each cell is a set of structures called chromosomes that carry all of the instructions (genes) for the cell to function. We generally have 23 pairs of chromosomes and inherit one of each pair from each parent. Sometimes a section of genetic material from one chromosome of a particular pair inserts into a chromosome from another pair. When the chromosome breaks do not pass through a gene and there is no gain or loss of material, it is called a balanced insertional translocation. Someone with a balanced insertional translocation (a carrier) usually has no health or developmental problems from this, although they may sometimes have difficulties when they want to have children.

The diagram below shows how breaks in two different chromosomes can result in a balanced insertional translocation.



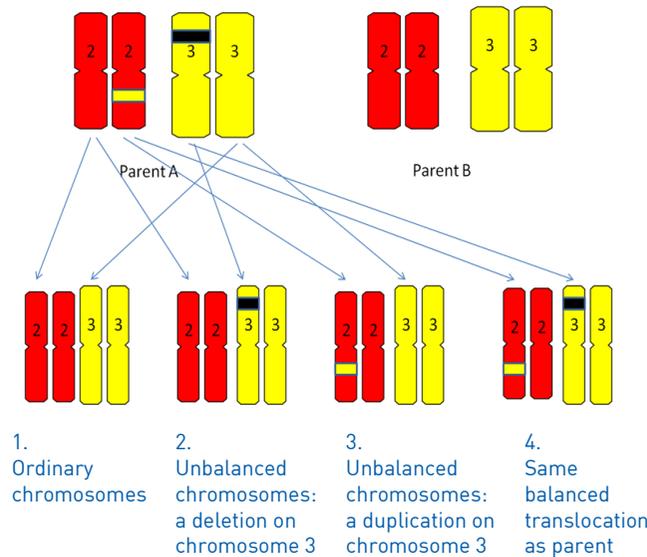
Most balanced insertional translocation carriers don't know that their chromosomes are any different to anyone else's. When they do find out, they have different questions about what the future holds for their family. Some people discover from a blood test when they have had a child with special needs or health problems caused by a chromosome disorder. Some people have repeated miscarriages or other fertility problems. Some people have a blood test as part of family investigations. Others find out by chance when they have a chromosome test for other reasons. Occasionally a balanced insertional translocation is found in a baby during pregnancy.

Why did it happen? Where did it start?

Balanced insertional translocations happen naturally. They are part of the natural evolution of species, including humans. Translocations can be new or they can be passed down in families from parent to child through the generations. New translocations occur when sperm or egg cells are forming or just after fertilisation.

What happens when someone with a balanced insertional translocation wants to have children?

When we have children, both parents will usually pass on one copy of each chromosome to their child. The same thing happens when one parent has a balanced insertional translocation. However, this can result in four different outcomes as shown in the diagram below.



Description of diagram: Parent A carries a balanced insertional translocation. Parent A passes on one copy of chromosome 2 (either the normal copy, or the copy with the insertion) and one copy of chromosome 3 (either the normal copy, or the copy with the deletion) to any offspring. Parent B is expected to pass on a usual set of chromosomes so that the resulting offspring has two copies of each chromosome. There are four possible outcomes from this.

Outcome 1 shows how a child can inherit a 'normal' set of chromosomes.

Outcomes 2 and 3 show how a balanced insertional translocation can be inherited in an unbalanced form. This means that there is some loss or gain of genetic material. This can cause failure to establish a recognised pregnancy, or apparent fertility problems due to repeated pregnancies with unbalanced chromosomes. A pregnancy with the unbalanced translocation could also miscarry or the baby could be stillborn. Alternatively, the pregnancy could continue to term and the child could have physical and/or learning problems.

Outcomes 4 The child can also inherit the same balanced insertional translocation as the parent. This should not cause any health problems in the child, although they may have difficulty later in life when they try to have children.

Ask your Geneticist or Genetic Counsellor.....

These outcomes will be more or less likely, depending on your particular translocation. Your genetic counsellor or geneticist should be able to give you more information about this.