What happens during pregnancy?

Some people choose to have testing during a pregnancy to find out whether the baby has inherited the unbalanced form of the translocation (meaning they have the incorrect amount of genetic information). This testing 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 have I got this translocation?

If you have been told you have a balanced insertional translocation, you may wonder 'Why me?' Remember, a *de novo* translocation probably arose when the egg or sperm cell, which was destined to create you, was formed. At the very latest, it arose in the earliest days of your mother's pregnancy. Everything that is known about balanced insertional translocations suggests that it occurs by chance. 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. Some people with a balanced insertional translocation may feel guilty. Some people who inherit a translocation find that the parent who passed it on feels guilty. Given that this occurs by chance, it is not a choice of an individual(s) and there is no reason for anyone to feel guilty.

Unique members with balanced insertional translocations

In October 2022, Unique had around 70 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 using the contact details listed.

Inform Network Support

Rare Chromosome Disorder Support Group
The Stables, Station Road West,
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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/donate
Please help us to help you!

Unique lists external message boards and websites in order to be helpful to families looking for information and support. This does not imply that we endorse their content or have any responsibility for it.
This guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed.

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Understanding Chromosome & Gene Disorders

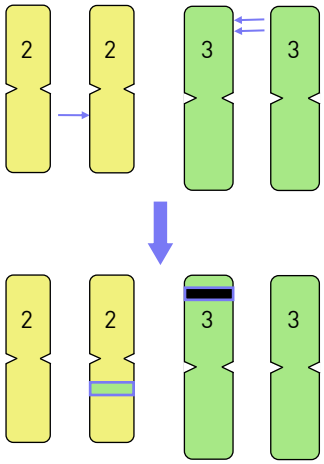
Balanced Insertional Translocations

rarechromo.org

What is a balanced insertional translocation?

Our bodies are made of trillions of cells. Our cells contain our genetic information which makes us who we are. To fit all of the genetic information into our cells, it is packaged into structures called chromosomes that carry all of the instructions (genes) that tell the body how to develop, grow and function. We typically have 23 pairs of chromosomes and inherit one of each pair from each biological parent. Sometimes a section of genetic material from one chromosome of a particular pair inserts into a chromosome from another pair. The point at which a chromosome has "broken" when a rearrangement such as this occurs is referred to as a breakpoint (see below for an illustration of this). When a chromosome break does 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 experience difficulty when they want to have children.

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



A. Two copies of chromosome 2 and two copies of chromosome 3. Arrows show the breakpoints

B. Part of chromosome 3 has inserted into chromosome 2, as shown by the green band. The black part in chromosome 3 shows where the genetic material has been lost from this chromosome.

Most balanced insertional translocation carriers do not know that their chromosomes are any different to anyone else's. When they do find out, they may have different questions about what this means and what the future holds for them and their family.

The ways that people may find out about the balanced insertional translocation include: discovery following investigations as their biological child requires special support or has health problems caused by a chromosome disorder. Some people may have repeated miscarriages or other fertility issues. Some people have a blood test as part of family investigations whilst 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 an ongoing 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 (*de novo* (dn)), or they can be passed down in families from parent to child through the generations. New translocations can occur when sperm or egg cells are forming or just after fertilisation (when the sperm and egg fuse together).

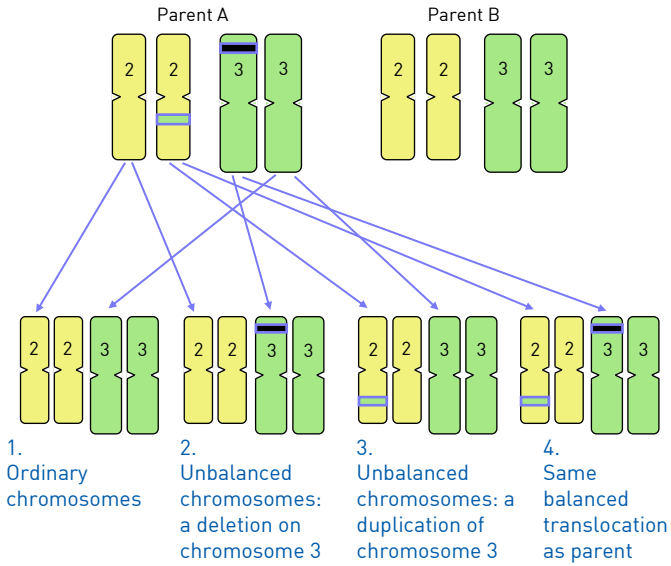
What happens when someone with a balanced insertional translocation wants to have biological children?

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 on the next page.

Ask your Geneticist or Genetic Counsellor.....

These outcomes will be more or less likely, depending on your particular translocation.

Your genetic counsellor or geneticist may be able to give you more information about this.



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.