Balanced Translocations
What is a balanced 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 in each pair from each parent. Sometimes a section from one chromosome of a particular pair changes places with a section from a chromosome of another pair. When the two breaks do not pass through a gene and there is no gain or loss of material when the chromosomes are looked at under a microscope, it is called a balanced translocation. Someone with a balanced translocation (a carrier) usually has no health or developmental problems, although they may sometimes have difficulties when they want to have children.

Most balanced 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 translocation is found in a baby during pregnancy. We talk about this situation on page 11.

This guide answers the most common questions that Unique members have asked. Of course it doesn’t replace your consultation with your genetics centre. Your genetic counsellor or geneticist is the best person to answer your questions about your personal situation.

Why did it happen? Where did it start?
Balanced 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. But even a family translocation was once a new one, though it may be generations back.

New translocations occur when sperm or egg cells are forming or just after fertilisation during the copying of the early cells that will become an embryo, then a fetus and then a baby. One study suggests that most new balanced translocations arise during sperm production and particularly in older fathers. They are not caused by men’s lifestyle, environment or work.
Your geneticist will offer to test other members of your family to find out whether your translocation is a new one or a family one. A small blood sample is needed for the test. Testing both your parents may be enough but if that is not possible, other family members may also be invited for testing.

**Why me? Is it my fault?**
If you’ve been told you have a balanced 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 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 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.

"Whenever I tell someone that I have a ‘chromosome disorder’ I feel like I’m telling them that I’m seriously ill."

"I just tell people that I have a problem that means I’m automatically in the high-risk group for screening in pregnancy."

**How common are balanced translocations?**
Around one person in 560 has a balanced translocation, so as a group they are not at all rare. Over the world, there are around 12 million balanced translocation carriers. When this guide was written, *Unique* had over 860 members with a balanced translocation. This may be the largest grouping anywhere in the world.

Any chromosome can make a swap with any other chromosome, anywhere along its length. This is what makes most specific translocations extremely rare, even unique. However, a few balanced translocations are found repeatedly. One is a translocation between chromosome 11 and chromosome 22 and is known as the 11;22 translocation and another is a translocation between the short arms of chromosomes 4 and 8. *Unique* has published separate information guides for these translocations.

**How common is my particular balanced translocation?**
Many translocations are so unusual that we can’t estimate how common they are. Whatever your translocation, your geneticist will research the medical literature to see if other families have been reported, although these may only be the tip of the iceberg. It’s quite likely that there will be other families with the translocation who have never been identified. Sometimes a genetic service does have other families with a particular translocation and sometimes *Unique* does. To find out if there are other families with the same translocation as you, ask your geneticist and/or *Unique*. 
Can we have children?
Most men and women with a balanced translocation can and do have children. There are usually four possible outcomes when a couple with a balanced translocation tries for a baby. These outcomes will be more likely or less likely, depending on your particular translocation. Your genetic counsellor or geneticist can help by saying which outcomes are more likely for you.

1. a child with an ordinary chromosome pattern
2. a child with the same balanced translocation as the parent
3. failure to establish a recognised pregnancy, or apparent fertility problems due to repeated pregnancies with unbalanced chromosomes
4. an ongoing pregnancy with unbalanced chromosomes which may not continue to term or may lead to the birth of a child with physical and/or learning problems

Some people wonder if having a balanced translocation makes other genetic disorders more likely in their children. There is no evidence at all of this.

The mother has a balanced translocation, inherited from her father (behind). After two miscarriages and two pregnancies terminated due to chromosome imbalance, she has had a daughter with the family translocation.

The father has a balanced translocation. This family had three miscarriages before having two daughters: one with the balanced translocation (left) and one with ordinary chromosomes (right).

“ There is a balanced translocation in all generations of my family which doesn’t have health implications for any of us. Out of three pregnancies, we have had a miscarriage of a baby with an unbalanced translocation, a child with a balanced translocation and a child with an unbalanced translocation who has special needs. ”
Having children: a family with a balanced translocation

Your geneticist can try to suggest which of these outcomes are more likely for your family.

Parents contribute one chromosome from each pair.

One parent with ordinary chromosomes

One parent with a balanced translocation

Either

1

Ordinary chromosomes

or

2

Same balanced translocation as parent

or

3 or 4

Unbalanced chromosomes: too much of one chromosome, too little of another

or

3 or 4

Unbalanced chromosomes: too little of one chromosome, too much of another

In this family, these chromosome arrangements should not cause health problems or special learning needs.

These arrangements with too much and too little chromosome material can lead to fertility problems, pregnancy loss or the birth of a child with special needs and sometimes health problems.
A child with the same balanced translocation as the parent: passing on a balanced translocation

If you or your partner pass the family balanced translocation on to one of your children, they should not have any new health problems or special needs caused by it.

When your children come to have their own children, they will be essentially in the same position as you were: they can have children with ordinary chromosomes; children with the same balanced translocation as themselves; fertility problems or pregnancies in which the baby has unbalanced chromosomes.

The tests available to them for detecting chromosome imbalances are likely to be available earlier in the pregnancy and more precise than when you were pregnant and generally waits for results will be shorter.

Any adult who has a balanced translocation can ask for an appointment with a genetics centre to find out what tests and services are available.

“ We feel infinitely more prepared than we were to support our own daughter (who has a balanced translocation like Mum) when she starts to think about having her own family. ”

Could I have a child with the family balanced translocation, but who has special needs?

In a family with an inherited balanced translocation, it would be unusual to have a child with special needs caused by the family translocation. The more people there are in your family with the balanced translocation and no special needs, the more confident you can be.

Very occasionally, it turns out that a translocation thought to be balanced in fact includes an unbalanced element too tiny to be seen under a microscope. New, more sensitive molecular ways of looking at chromosomes such as microarrays can help to detect tiny pieces of missing or extra chromosome material. In this extremely rare situation, the carrier may have some special needs.
Will men and women with a balanced translocation have fertility problems?
Most men and women with a balanced translocation are able to have children. It may take longer for a pregnancy to be established because a proportion of their sperm or eggs have unbalanced chromosomes and any embryos cannot survive.

Miscarriage and pregnancy loss
It’s surprisingly common to miscarry, often even before a woman knows she is pregnant. When anyone discovers they are pregnant, there is a one in seven possibility that the pregnancy will end in miscarriage. Usually, no obvious reason is found - which is why doctors do not normally investigate until after the second or third loss.

When one member of the couple has a balanced translocation, the possibility of losing the pregnancy, usually in the first three months but sometimes later, rises to perhaps 20-30 per cent and is occasionally over 50 per cent. Miscarriages happen in this situation because the developing baby has unbalanced chromosomes - too much chromosome material or too little. The greater the imbalance, the more likely it is that pregnancy will not be established in the first place or, if it is, it will be lost.

Each time you try for a pregnancy, the possibilities are the same - of a baby with normal or balanced chromosomes, or of a baby with unbalanced chromosomes.

If you have many repeated miscarriages without any successful pregnancies, you may want to discuss this with your genetic counsellor or geneticist. Questions that you may want answered include knowing whether a baby with unbalanced chromosomes can survive to birth, whether they may die soon after birth and if they survive how severely they will be affected mentally and physically. If you know that you have a family balanced translocation inherited from one of your parents, you have reassuring proof that it is possible to have a healthy child.

When you have a miscarriage you may be invited to send the ‘products’ for genetic testing. This is a hard decision to make at a traumatic time but it can help to build up a picture of what is going on, and can also help to further genetic knowledge.

“It was only coming to the Unique conference and meeting other people with balanced translocations that gave me the courage to try again for a pregnancy. I have now had nine miscarriages - and two healthy and very beautiful children!”
You’re pregnant: how soon can you find out about the baby’s chromosomes?
The earliest test that will currently show your baby’s chromosome make-up during pregnancy is based on chorionic villus sampling (CVS) at about 11-13 weeks. A fine needle removes some tissue from the placenta (afterbirth). You will usually be given the results in three days to two weeks. The test itself may make it slightly (about 1 per cent) more likely that you will miscarry, and if this happens, it will occur within a few days of the test.

Your baby’s chromosomes can also be analysed from cells obtained at amniocentesis. A fine needle removes fluid from the water surrounding the fetus in the uterus. This is performed at about 16 weeks of pregnancy and you will usually have confirmation of the baby’s chromosome make-up in 10-14 days. Amniocentesis is associated with a slightly raised possibility of miscarriage of about 0.5-1 per cent.

These tests can show whether your baby has too much or too little chromosome material, but they cannot tell you how your baby will be affected. A child with unbalanced chromosomes is very likely to have special needs.

A fetal anomaly scan can help to show if the baby is likely to have major health concerns by revealing structural problems with the heart or other major organs. But a full picture of how a baby with unbalanced chromosomes is affected can only be drawn after birth.

You should have the opportunity to talk over what is involved in these tests with your midwife or obstetric doctor and they should refer you to a genetic counsellor or geneticist if you want more information about any abnormal results.

If these tests give an abnormal result, you will have the choice of continuing the pregnancy and preparing yourselves for the birth of a child with special needs or terminating the pregnancy. Many Unique members have faced these decisions.

“Terminating the pregnancy was an agonising decision for my husband and me but it was easier knowing that other couples have made the same decision.”

Other reproductive choices
You may want to look at other reproductive choices. What is available and whether you have to pay depends on what country you live in, but can include the following options.

Pre-implantation genetic diagnosis (PGD)
Pre-implantation genetic diagnosis (PGD) gives the opportunity to select an embryo without an unbalanced translocation that can be placed back in the
womb. The procedure uses *in vitro* fertilisation (IVF) techniques, involves checking the chromosomes of 3-day-old embryos and only transferring those with normal or balanced chromosomes to the womb. The method used at the moment does not distinguish between embryos with normal chromosomes and those with a balanced translocation.

*Unique* has members who have used PGD to have healthy children and are happy to share their experiences. The availability of PGD varies widely between countries and it is not available for chromosome translocations in all clinics offering PGD. Success rates are 20 per cent per cycle started and 30 per cent per embryo transferred (take home baby rate). In the UK the procedure is expensive but 85 per cent of couples get funding. Some people find it emotionally demanding and counselling and support are vital before and throughout the process. If you want to find out more about your options for PGD, ask first for a referral to a genetics centre.

**Intracytoplasmic sperm injection (ICSI)**
Where there are concerns over fertility, there are ways to select active, fast-moving sperm with sperm analysis and use them in intrauterine insemination or in intracytoplasmic injection (ICSI). In ICSI a single sperm is pre-selected. It is then injected directly into an egg that has been ripened and removed from the mother in the same way as if she was having *in vitro* fertilisation (IVF). This procedure can be combined with PGD to ensure that only embryos with balanced or normal chromosomes are transferred to the womb.

**PGD process**
Mother given fertility drugs to stimulate ovaries → multiple eggs produced and collected → eggs fertilised by IVF to produce embryos → embryos develop for 3 days, then 1 cell removed from 6-10 cell embryo and analysed for unbalanced translocation → 1 or 2 embryos with balanced translocation/ ordinary chromosomes transferred to uterus → 10-12 days later pregnancy test → if pregnancy is established, prenatal diagnosis using CVS or amniocentesis is offered.

**Egg or sperm donation**
Couples may consider egg or sperm donation if they are not successful in having a healthy child. Donated sperm can be given using intrauterine insemination (IUI) following a woman’s natural cycles or after stimulating ovulation with medication. Donated sperm or eggs can come from someone you know or be supplied by a clinic. The donated egg is combined with the partner’s fresh sperm and once the recipient’s womb has been prepared with hormones, the embryo is transferred to the womb using a normal IVF procedure.
Adoption

Some families choose to adopt children.

“After six miscarriages, my husband and I decided to look at adoption. We are now proud parents of two beautiful (adopted) children and have never looked back. Adoption is a way of giving children a loving family who may not be so lucky otherwise. An unconceived child does not need to be created or yet have the right to live. A child who has been born does.”

Your children: More questions

Your other children

Your other children can have their chromosomes tested once they are old enough to decide that they want to know. If you feel strongly that you want to know their chromosome status but they are still too young to decide for themselves, talk it over with your genetics service. The guidance about when it is best to test and tell is flexible and intended to be in families’ best interests. If your child is developing normally, it is most likely that they will either have normal chromosomes or the family balanced translocation. A normally developing girl or boy with the family balanced translocation can expect to be unaffected by it until they have children of their own.

When and how should we tell a child about their balanced translocation?

Unique is preparing a separate information guide to telling children about having a balanced translocation.

We have one child with special needs. If we have another child with special needs, will they be affected in the same way?

If you have another child with unbalanced chromosomes, it is very likely that they will have special needs. But their needs won’t be exactly the same as their brother or sister’s. Depending on your translocation, they may have a different unbalanced chromosome make-up or they may have the same make-up as your older child. Even if the two children have the same chromosome make-up, the effects of having unbalanced chromosomes can vary quite a lot - just as brothers and sisters with ordinary chromosomes are different from each other.
Will my balanced translocation affect my health?
For the great majority of the millions of people with a balanced translocation there is no effect on their own health. For any child who inherits the family balanced translocation there should also be no health problems caused by the translocation. Balanced translocations don’t usually have any effect on health or development because in the great majority of cases the points in the genome where chromosomes have broken and re-joined don’t disrupt the smooth running of the genes.

Can a balanced translocation be cured?
No, it can’t. But a balanced translocation is very unlikely to cause any problems until the carrier wants to have children.

A balanced translocation is found in the baby during pregnancy
Occasionally a baby is found to have a balanced translocation at chorionic villus sampling or at amniocentesis. First the parents’ chromosomes should be checked to see if the translocation is inherited. If it is inherited, and the parent has no problems from the translocation, it’s unlikely that the baby will have any problems either.

If both parents have normal chromosomes, the baby’s translocation is a new one (de novo). In most cases, the baby will be unaffected. But there is a risk that the chromosome breaks have disrupted important genes or that the break points are not as clean as they appear. The mother should be offered high-level ultrasound scans to look for any physical abnormalities. Couples should also have very early access to genetic counselling to identify any possible problems.

It is sometimes helpful to analyse the chromosomes again using a sensitive molecular or DNA technology that will show whether particular genes have been left out, added in or disrupted at the break points.
There is a closed Facebook group for families affected by balanced chromosome translocations at www.facebook.com/groups/35507179052

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 information 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. It was compiled by Unique in collaboration with Professor Maj Hultén BSc, PhD, MD, FRCPath, formerly Professor of Reproductive Genetics, University of Warwick, UK 2009.

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