



Understanding Chromosome & Gene Disorders

Triploidy

Triploidy

Triploidy is a disorder that arises at conception when a baby starts life in the womb with a complete extra set of chromosomes. Chromosomes are the microscopically small structures in the nucleus of the body's cells that carry genetic information. Most people have two sets of 23 chromosomes, making 46 in all. Babies who have too many chromosomes or too few are at increased risk of developmental problems and birth defects. Babies with triploidy have an entire extra set of chromosomes, making 69 in all. Most studies seem to suggest that around two thirds of triploid pregnancies are boys, while around one third are girls.

Triploidy is not the same as trisomy. People with trisomy have a single extra chromosome, making a total of 47. Down's syndrome, where there is an extra chromosome 21, is the best known form of trisomy.

Can a baby live with triploidy?

Very occasionally, babies with triploidy are born and live for a few hours, days or weeks. One baby reported in the medical literature lived to 10½ months, but this is very rare. Two out of three pregnancies miscarry in the first trimester and almost all other babies die later or are stillborn.

Is triploidy rare?

At conception, triploidy is thought to affect an estimated one to three per cent of confirmed pregnancies.

Why did this happen?

In the great majority of families, having a triploid baby is almost certainly a one-off event. Nothing that parents did caused it and there was nothing they could have done to prevent it, either. No studies have identified any clear risk factors that make triploidy more likely. Certainly, older mothers and older fathers are no more likely to have an affected baby than younger parents.

Triploidy can occur both with normal reproduction and after assisted reproduction. However a baby is conceived, at conception it is a very common chromosome disorder.

What happens in triploidy?

A baby boy or girl with triploidy starts life in one of three ways.

- The egg is fertilised by a sperm with a double set of chromosomes (diandry)
- The egg is fertilised by two sperm (dispermy, a type of diandry)

In both forms of diandry, the fetus has two sets of chromosomes from the father and a single set of the mother's chromosomes. Diandric pregnancies tend to have a large placenta. The baby is usually a good size but is more likely to miscarry during the first trimester. See [Partial molar pregnancies](#) (page 6)

- The egg-producing cells fail to divide properly and so the mature egg contains a double set of chromosomes (digyny). The baby has two sets of

maternal chromosomes and a single set of paternal chromosomes. Digynic pregnancies have a small placenta and the baby and particularly the body is usually severely growth restricted. Compared with diandric pregnancies, more babies with digyny live into the second trimester and studies have shown that four out of five babies who lived for more than a month after birth were digynic.

How is triploidy detected?

In early pregnancy

Many babies are only found to be triploid after they, die, sadly, in the womb. Others are first found by routine pregnancy scans and maternal serum screening tests. These tests do not provide a firm diagnosis of triploidy, but they do suggest the need for more detailed investigation.

Tests offered vary between hospitals, areas and countries but these tests have detected early signs of a triploid pregnancy and shown differences between a diandric pregnancy and a digynic pregnancy.

■ Diandric pregnancy

By the time an early pregnancy scan is carried out at 10-14 weeks, the baby is often already on the small side and the pregnancy may be re-dated. The scan will show an unusually large placenta and a baby of the expected size for the gestation. Typically, the nuchal pad (at the back of the neck) is thick.

The first trimester serum screening blood tests will usually show high free beta hCG (human chorionic gonadotrophin) and AFP (alpha fetoprotein) levels, while PAPP-A (pregnancy associated plasma protein-A) levels are low.

■ Digynic pregnancy

The early pregnancy scan will show a small placenta and a very small baby with a particularly small body. The results of serum screening tests may be different to a diandric pregnancy: typically, hCG and PAPP-A are low and AFP levels are low to normal.

In mid-pregnancy

Two possible results from a mid-pregnancy serum screening test might suggest a triploid pregnancy.

AFP (Alpha Fetoprotein)	hCG (human chorionic Gonadotrophin)	Oestriol	
Low/normal	Low	Low Possible	Possible triploidy
High	High	Low/normal	Possible triploidy

The mid-pregnancy anomaly scan is likely to show similar changes to the early pregnancy scan. In a digynic pregnancy, there may also be very little amniotic fluid. Apart from being small, the baby may appear quite unharmed but some anomalies may be visible.

Diagnosing a triploid pregnancy

If scans and blood tests raise concern, you will be offered a detailed scan at a specialist referral centre. If this confirms the unusual findings, you will be offered a test to diagnose triploidy by examining a tiny sample of cells from the developing placenta or from the amniotic fluid surrounding the baby.

Chorionic villus sampling (CVS) A sample of the developing placenta is removed either through a fine needle passed through the abdomen and the wall of the uterus or through a catheter passed through the cervix.

Amniocentesis A hollow needle is passed into the sac around the baby and a small quantity of amniotic fluid is removed. The amniotic fluid contains cells shed by the fetus. Under a microscope, the extra chromosomes are clearly visible.

It can take about two weeks for results of these tests to come through but most hospitals offer at least one way to get a result within two to three days.

Making a decision

The decision whether or not to continue your pregnancy is complex and personal. The placenta may have particular changes that suggest to medical staff that you may develop pre-eclampsia in mid-pregnancy. As this is a serious complication that can be life-threatening, it is a factor to consider when making this very difficult decision.

People can feel very isolated and be aware that hospital staff may know little or nothing about triploidy. Most hospitals should be able to arrange a consultation with a genetic service where a counsellor or geneticist will have expert knowledge of triploidy.

Whatever choice couples or individuals make, they need warmth, support and practical information. In the UK, the charity ARC (Antenatal Results and Choices) exists to support people facing these agonising decisions. ARC can offer various levels of support, including personal support by a trained volunteer who has experienced a triploid pregnancy. The *ARC Parents' Handbook*, with information on the practical and emotional issues, should be given to couples by their hospital. If this does not happen, they should contact ARC (see [Sources of support](#) on page 7).

Miscarriage

The great majority of triploid pregnancies end in miscarriage. If you start to bleed or feel pains similar to labour pains, you should contact your hospital. You will normally go into hospital and whether the miscarriage takes place there or at home, you will usually have an evacuation under anaesthetic afterwards to ensure that all the pregnancy tissue has been cleared from your womb.

Stillbirth

Occasionally, a triploid pregnancy continues past week 24 into the third trimester. Despite vigilant antenatal care, it is likely that your baby will die in

the womb. You will then probably have an induction and your baby will be born after going through labour. This is emotionally harrowing and you may have conflicting feelings about your baby afterwards. Some parents find comfort in spending time with their baby, keeping treasured mementoes and photographs and in arranging a funeral. But no one responds in exactly the same way.

Continuing the pregnancy

Some people wish to allow the pregnancy to continue or learn about the triploidy when it is no longer possible to make a choice. While it is true that most babies with triploidy die before birth, some are born alive and live for a few hours, days or weeks. A few live for a few months.

There is support available in the ARC handbook (see page 4) or on their website. People who visit their website at www.arc-uk.org should click onto For Parents, then Making Decisions and Continuing a Pregnancy.

People who choose to continue their pregnancy have said that they value it particularly when medical staff treat the pregnancy as normal, if high-risk. Their baby is scanned and the heartbeat checked regularly. If the mother is continuing the pregnancy despite the possibility of developing pre-eclampsia, her blood pressure and urine protein are meticulously monitored. If pre-eclampsia develops, labour will be induced.

Birth

So long as triploidy has been diagnosed before birth, couples can expect a normal delivery. Research reports suggest that many newborn babies have respiratory difficulties but parents' experience shows that some babies are born breathing well, while others need supplemental oxygen for a short time. Some babies without any apparent health problems may be able to feed, although their sucking tends to be weak.

Parents given a chance to hold their baby after birth have commented how normal he or she appears. The baby may have unusual features such as joined fingers or toes and may be small but overall may look quite like any fragile new baby.

Living with a baby with triploidy

It is hard to prepare for the experience of living with a baby with triploidy. Most babies live for hours or days, but occasionally they live longer. The medical literature states that the longest known survivor lived for 10½ months. This little boy was able to go home and medical staff noted that he smiled and appeared to recognise his parents and brother. Eventually, he died after a seizure. Of the babies pictured (right) one lived for a few days, one for 20 days.



Ending a pregnancy with a triploid baby

Recent studies suggest that the majority of people choose to end their triploid pregnancy. Uncertain couples may wish to contact Antenatal Results and Choices or to log on to a website at <http://www.aheartbreakingchoice.com> that exists to support people who face the choice of whether or not to continue a pregnancy where the baby is likely to be severely affected.

Women who are not yet 14 weeks pregnant are usually offered a D&C (dilatation and curettage, also called D&E – evacuation) or a suction curettage under a general anaesthetic. Once you have passed 14 weeks, you usually go through labour following a prostaglandin induction. In some hospitals it may be possible for you to be offered a general anaesthetic.

Partial molar pregnancies

It is important to establish which parent contributed the extra chromosomes to the pregnancy because of the link with a partial molar pregnancy. In a partial molar pregnancy, the placenta grows very large and cyst-like, until it looks like a bunch of grapes. Partial molar pregnancies are not found when the extra chromosomes are from the mother, but they may be found when the chromosomes are paternal (diandric).

Women who have had a molar pregnancy can expect to be followed up to ensure that there is no abnormal tissue left. Remnants of mole tissue can grow and carry a very small but identifiable risk of developing into a form of cancer called choriocarcinoma. This is treatable and the aim of follow-up is to ensure that if it does develop it is caught and treated as early as possible. Follow-up is usually for six months to two years.

Trying again

There is no right length of time to wait before trying for another baby. However, after a partial molar pregnancy, women will usually be advised to wait until follow-up is complete. Women who are followed up for two years are usually advised to wait until they have had normal blood and urine test results for six months.

Can it happen again?

Triploidy is usually a one-off event and the chances of it happening again are no higher than for anyone else in the population. There are some extremely rare families who have had more than one triploid pregnancy and some women with experience of more than one triploid pregnancy have posted messages on the triploidy message board. However, as triploidy is so common at conception, it is most likely that these repeat pregnancies with triploidy occurred by chance.

Sources of support

If you would like email contact with someone else with experience of triploidy, a mother who formerly moderated a triploidy loss webgroup is happy to answer questions. Email farmerswife1991@hotmail.com

Unique also has members with different experiences of triploidy. Please contact us if you would like to be put in touch with others.

ARC Antenatal Results and Choices

www.arc-uk.org

345 City Road

London EC1V 1LR

United Kingdom

Helpline: +44 (0) 845 077 2290

Or (0) 207 713 7486 via mobile

info@arc-uk.org

Stillbirth & Neonatal Death Society

www.uk-sands.org

28 Portland Place

London W1B 1LY

United Kingdom

Helpline +44 (0) 20 7436 5881

support@uk-sands.org

Unique mentions other organisations' message boards and websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it.

Support and Information



Understanding Chromosome & Gene Disorders

Rare Chromosome Disorder Support Group,

The Stables, Station Road West, Oxted, Surrey RH8 9EE, United Kingdom

Tel: +44(0)1883 723356

info@rarechromo.org | www.rarechromo.org

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!

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. It was compiled by Unique and reviewed by Dr Tessa Homfray, consultant clinical geneticist, St George's Hospital, London 2004 and by Professor Maj Hulten BSc, PhD, MD, FRCPath, Professor of Medical Genetics, University of Warwick, UK. 2005.

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