

Families say ...

'David challenges us to give beyond ourselves and is a thermometer of our selfishness. We value simplicity and live knowing that life is a precious gift. We don't value things as precious commodities but each other as our greatest resources. David makes us budget our energy and live flexibly.'

Inform Network Support



Rare Chromosome Disorder Support Group,
G1 The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

Facebook groups

Diploid Triploid Mosaic Syndrome (or Mosaic Triploidy)
www.facebook.com/groups/111944605491961
Diploid Triploid Mosaic Europe
www.facebook.com/pages/Diploid-Triploid-Mosaic-Europe/863765023635648

When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a full leaflet about the effects of Diploidy Triploidy.

This information sheet 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. The information is believed to be the best available at the time of publication and the medical content has been verified by Dr Jacques Giltay, Clinical Geneticist, University Medical Center, Utrecht, The Netherlands and by Professor Maj Hulten, Professor of Medical Genetics, Warwick University, Unique's Chief Medical Adviser 2004.

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Rare Chromosome Disorder Support Group
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Why?

Most chromosome disorders arise out of the blue from an event that occurred when the sperm and egg cells were forming or very soon after conception (when a baby is made). As a parent there is nothing you can do to control this and no environmental, dietary or lifestyle factors are known to cause diploidy triploidy. There is nothing you did before you were pregnant or during pregnancy to cause this and nothing you could have done to prevent it.

The extra set of chromosomes comes either from the father or the mother and may have been incorporated either when two sperm fertilised a single egg or because the egg from the mother contained two copies of each chromosome instead of the usual single copy of each. The fertilised egg - which would be triploid after conception - may then have tried to 'self correct' with only partial success, leading to the two different types of cell, both diploid and triploid.

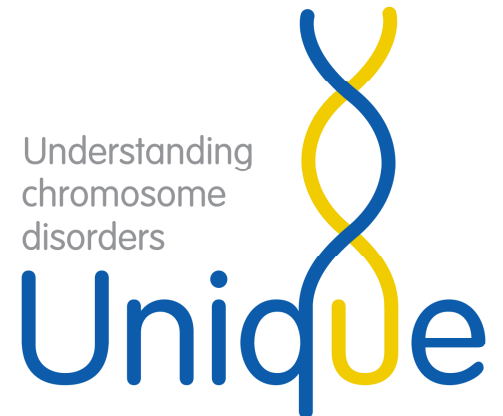
The extra chromosomes can occasionally first be incorporated after conception – but the effects on the baby are similar.

Can this happen again?

Diploidy triploidy is almost certainly a one-off event and the chances of it happening again are generally no higher than if you never had an affected baby. There are no medical reports or records at *Unique* of couples having more than one diploid triploid pregnancy. However, it is natural to be concerned and couples who want it may be offered prenatal diagnosis in their next pregnancy.

What is life expectancy?

Your child's doctors are in the best position to answer this difficult question. *Unique's* oldest member is 20 years old and the oldest person described in a medical text is 21 but there must certainly be older people with diploidy triploidy.



Diploid Triploid children

rarechromo.org

What is Diploidy Triploidy?

A baby with diploidy triploidy has too many chromosomes in some of the cells of the body. People usually have 46 chromosomes in every cell (apart from sperm and eggs which have 23 chromosomes each).

A baby with diploidy triploidy has a mixture of cells - some with the standard number of 46 chromosomes (called **diploid**) and some with an entire extra set of chromosomes, making 69 in all (called **triploid**).

When a baby is made at conception, the father's sperm usually contributes a set of 23 chromosomes and the mother's egg also contributes a set of 23 chromosomes. In a pregnancy with full triploidy, there is an extra set of chromosomes in each cell that has either come from the mother or from the father. In a baby with diploidy triploidy, cells with 46 chromosomes exist alongside cells with 69 chromosomes.

Chromosomes contain the genes that give instructions, determining the way the body develops and works. When there are too many, some parts of the body develop as you would expect, but others do not.

Triploid cells aren't the same as the cells in a baby with trisomy. Babies with trisomy have a single extra chromosome – 47 instead of 46. The most common trisomy causes Down's syndrome.

What is the link between the number of triploid cells and the effect on the child?

There is no obvious link. The link between the level of learning difficulty and the ratio of triploid to diploid cells is not clear and is not fully understood.

Development

Every child with diploidy triploidy is unique. Yet there are common features.

■ Physical development

Physically, children tend to be small. They are often small at birth and never completely catch up. A growth or hormone specialist can advise whether injections of growth hormone will increase your child's eventual adult height.

One side of the body may be larger than the other. This may be subtle or so obvious that your child takes different shoe sizes. Any facial asymmetry may well become less obvious during childhood.

Around the age of 4, children tend to put on weight around their waist, so they appear to have a fat chest and stomach.

Some children have streaks or patches of irregular skin colour, with areas of lighter colour mingling with patches of darker skin.

Other unusual physical signs, such as joined or webbed fingers and toes, incurved fingers and low-set ears with an unusual shape, are also found in many children with other chromosome disorders.

■ Learning

Most children are expected to have some degree of learning difficulty. The severity is hard to predict and the eventual range is very wide, but cases reported in the medical literature and *Unique's* experience suggest that almost all children will need support with formal learning. With supportive speech and language therapy most children will learn to say some words and some will become fluent speakers.

■ Behaviour

No behaviour problems have been specifically identified with diploidy triploidy. *Unique's* evidence is limited but it suggests that children are calm and loving.

Medical conditions

- **Seizures** affect around half of children described in the medical literature. They often start in babyhood or early childhood and can usually be controlled effectively with anti-epileptic medicines.
- **Heart conditions** such as holes between the chambers of the heart (atrial septal defects - ASDs and ventricular septal defects - VSDs) occur more often than in other children.
- **Hypotonia** (low muscle tone, floppiness) is common. Babies tend to reach their milestones late but in general can be expected to learn to walk. The hypotonia usually improves with age.
- **Boys** may be born with a small penis and undescended testes. An operation (orchidopexy) can be carried out during childhood to retrieve the testes and fix them in the scrotum.
- **Precocious puberty** appears to affect some girls, with early signs sometimes starting in babies. Hormone treatment can be given but symptoms may disappear naturally.
- **Kidney** problems have been described and babies can expect to have their kidneys and urinary tract examined.

How is diploidy triploidy detected?

In pregnancy, your baby's small size or the results of serum screening tests may cause concern. You will then be offered a chorionic villus test or amniocentesis to examine cells with the same chromosomes as the baby. Cells from the developing placenta or the amniotic fluid are treated so that the chromosomes can be examined under a microscope. The extra chromosomes are easily visible.

After birth, your baby's appearance, behaviour or development may suggest the need for a chromosome analysis. Cells may be taken from a skin biopsy as well as from a blood sample because the number of cells with extra chromosomes can vary considerably. In more than three quarters of children with diploidy triploidy, all blood cells are diploid (they have 46 chromosomes), while skin cells are more likely to be triploid (and have 69 chromosomes).