Why?

Nothing that parents did caused their child to have T8M and nor could it have been prevented. It is usually caused by a mistake in cell division very soon after conception. T8M is different in this respect from other trisomy chromosome disorders such as Down's syndrome (trisomy 21) which arise most often from a system fault in the creation of the egg and are more common in older mothers. However, it is true that T8M is slightly more common in older parents.

Can this happen again?

Trisomy 8 mosaicism almost always occurs completely by chance. There are no reports in the medical literature of families with more than one affected child and none are known to *Unique*.

Prenatal diagnosis

Prenatal diagnosis of any chromosome disorder involving mosaicism is extremely difficult. It requires a combination of tests because of the varying proportions of cells with an extra chromosome and cells with normal chromosomes in different parts of the body. In particular, a positive T8M result on chorionic villus sampling (CVS) does not necessarily mean that your baby is affected. In most pregnancies where T8M is found on CVS, the extra chromosome is only found in the developing placenta and not in the baby. Even amniocentesis alone is not an accurate guide to T8M. The most accurate diagnosis comes from combining the results of an amniocentesis with high resolution ultrasound scans and ideally a direct sample of the baby's blood cells taken from the umbilical cord.

One mother had identical (monozygotic) twins, both with trisomy 8 mosaicism, but each showing quite different effects of the extra chromosome.

Families say …

When S is well he is a delight – he has the most infectious laugh and smiles. Having S has empowered us, his parents, to re-evaluate our lives and surprisingly we have done more since having him - age 8.

The pleasure when he achieves something is priceless - age 14.

S is a great encourager and comforter if you're ill or feeling down. He has a fantastic sense of humour and comes out with amazing one-liners ! - age 20.

Inform Network Support

Understanding chromosome disorders Unique

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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 Mosaic Trisomy 8.

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 has been reviewed by Dr Jill Clayton-Smith, consultant clinical geneticist, St Mary's Hospital, Manchester and by Unique's Chief Medical Adviser 2004.

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Trisomy 8 Mosaicism

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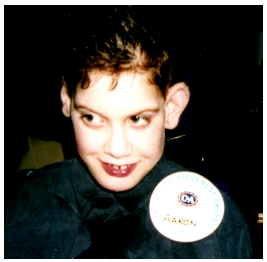

What is trisomy 8 mosaicism?

People usually have 46 chromosomes in each cell in their body. These chromosomes are small packages of genes or instructions that tell the body how to develop and work.

Trisomy 8 mosaicism (T8M) is caused by having a mixture of cells with an extra chromosome 8 and cells with the standard number of chromosomes. Your geneticist will tell you what proportion of cells have the extra chromosome but there is no clear link between this number and the effects of T8M on your child.

T8M can have a huge range of effects – from nothing remarkable to distinct physical features and effects on development. The extraordinary variability of T8M together with your child's personality and other genes will mean that he or she is unique.

The features of T8M are extremely variable. Some people appear completely unaffected, either physically or developmentally. Others are born with a variety of physical features and experience a range of developmental delay. This list of 'typical' features will inevitably mention some that do not affect your child.

You may notice some of these typical features:

- Deep lengthwise skin creases in the soles and sometimes palms
- Stiff or restricted movement in some joints
- Slim body with narrow chest, shoulders and pelvis
- Well-formed, pear-shaped nose; somewhat pouting lower lip
- Large ears with a thick external ridge.

Many babies are a normal length and weight at birth.

How common is T8M?

Diagnosed T8M is uncommon, affecting only one in every 25,000 to 50,000 babies. In reality, the condition may well be more common than this as people with no obvious signs will not be investigated. For reasons that are not understood, T8M is three to five times more common in boys than in girls. Fifty families belong to *Unique* and contact with other families can be arranged.

How can T8M affect a child's development?

- **Motor development**
  The range of achievement is extremely broad, but a typical child will be somewhat delayed in reaching developmental milestones, sitting for example around 8 months and walking between 18 months and two years.
- **Learning**
  The range is huge, from no effect at all to severe learning difficulties. As far as learning is concerned, your child will 'write his own book'. His personal ability to learn will become quite apparent within the first three years of age.
- **Speech**
  People often say that speech is a weak point in children with T8M. Children may have articulation problems and their understanding is usually ahead of their ability to speak. With speech therapy, most youngsters have made great progress by adolescence.

How is T8M detected?

T8M is sometimes discovered during pregnancy, usually when testing for other more common conditions. But it is most commonly detected after birth when doctors notice a baby's unusual features or development and request a chromosome analysis. This will show varying levels of T8M cells, frequently more in the skin than in blood samples.

Medical concerns

- **Joints** are typically clenched, tight or do not open completely. Large joints (such as the hips and shoulders) can be involved as much as small joints (such as wrists, ankles and fingers). Some joints are not properly formed or the bones are fused.
- **Heart conditions** are common, so your baby will be carefully monitored. Monitoring may be all that is needed because in time many heart conditions heal on their own. However, some babies need medicines or surgery.
- **Urinary and kidney conditions** are also common so your baby would normally have an ultrasound scan of the urinary system.
- Some babies have some **bones** that are unusually shaped or missing bones, in the knee cap, for instance. Others have extra bones, such as an extra pair of ribs. This does not usually affect them at all but the paediatrician will want the whole skeleton carefully to be carefully checked. Occasionally **vertebrae in the spine** fuse or are not completely formed.
- Some boys are born with **undescended testicles**. The hole that is usually at the end of the penis may be on the underside instead (**hypospadias**). Both of these conditions can be corrected with a straightforward surgical operation, usually before school age.
- Quite a few children develop an **eye condition** or are born with one. Some conditions do not affect vision at all but others, like a squint (strabismus) or cataracts do and your baby will need treatment and possibly surgery.
- As children grow, their spine may develop a sideways or outward curve, known as **scoliosis** or **kyphosis**. All that may be needed is exercise or physiotherapy, but some children are helped with special supportive clothing and seats.