Why did this happen?

The causes of chromosome disorders such as T9M are not yet fully understood but it is known that there is nothing you did before you were pregnant or during pregnancy that caused it and also nothing you could have done to prevent it.

A trisomy can occur when there is a mistake in the natural process by which chromosomes copy themselves and separate during cell division. This may happen when a mother's eggs are forming. These mistakes are more common in older mothers, but most babies with a trisomy are still born to younger mothers. An attempt very soon after conception (when a baby is made) to correct the mistake can lead to the presence of the different cell lines if the fault is corrected in some cells and not in others. T9M can also arise for the first time after conception when the fertilised egg is dividing and multiplying.

Can it happen again?

When both parents have normal chromosome patterns the chances of having another child with T9M are hardly any higher than for the rest of the population. Occasionally one parent has a rearrangement of one of their own chromosome 9s that does not affect them but may make a chromosome disorder in a future pregnancy more likely. We would recommend that you discuss this with your clinical geneticist who can advise you about your own individual situation.

Prenatal diagnosis

Diagnosing a trisomy mosaicism during pregnancy is complex. Sometimes more than one chromosome study is needed as well as detailed ultrasound scans to look for any effects on the baby's body systems and growth. Your obstetrician will give you as much certainty as possible and support a rapid referral to the clinical genetics service for further advice if you wish.

Families say …

" She has totally overwhelmed us all both in what love and total trust she has for us but also in the capacity in which we can give the same back to her.

" She is loving, always smiling, has a great sense of humour. She keeps us right.

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 trisomy 9 mosaicism.

This leaflet 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 verified by Dr Jill Clayton-Smith, Consultant in Clinical Genetics, St Mary's Hospital, Manchester 2005 and by *Unique*'s chief medical advisor 2004.

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Trisomy 9 mosaicism

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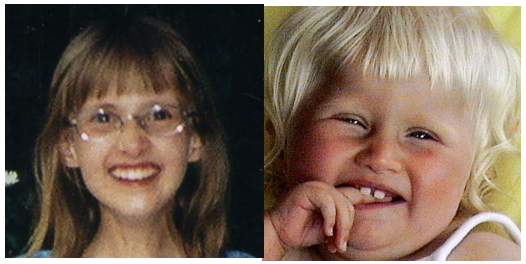


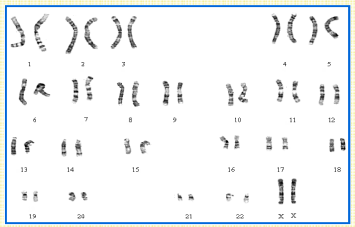
What is trisomy 9 mosaicism?

Trisomy 9 mosaicism (T9M) is a very rare chromosome condition. It may be detected during pregnancy, when a baby is born or in an older child being investigated for a medical condition or delay with development.
People with this rare condition have an extra chromosome 9 (known as a trisomy) in some of the cells in their body.

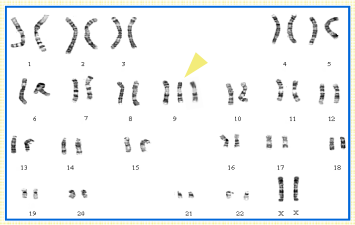
Chromosomes carry the instructions (genes) that tell the body how to develop and work properly, so having one extra makes it more likely that a baby will have problems with growth and development.

In T9M, as well as cells with the extra chromosome 9, there are cells with the usual number of 46 chromosomes. These cells generally lessen the impact of the chromosome disorder, but having cells with different chromosome counts (mosaicism) makes it more difficult to predict the outlook.

T9M occurs in both boys and girls, but appears to be around three times more common in girls.



Normal female karyotype - 46,XX



Female karyotype with an extra chromosome 9 - 47,XX,+9

Medical concerns

Some children have no medical problems at all. When problems do occur, the following are the commonest.

Feeding difficulties

Babies tend to suck weakly and may have difficulty coordinating sucking with swallowing. Their feeds may return readily up the food pipe (gastro oesophageal reflux). Reflux carries a risk of causing chest infections known as aspiration pneumonia. Careful positioning for feeds, prescribed antacid medicines to soothe an inflamed oesophagus and feed thickeners to help keep liquids down usually controls this.

Small size and slow weight gain

Babies are typically small at birth and find it hard to gain weight. High calorie feed supplements help but some children need to be tube fed for a time and a small number benefit from gastrostomy (tube) feeding direct into the stomach.

Lax joints

Hip dislocations and club foot (talipes) are fairly common. Treatment usually involves some weeks or months in a brace or plaster and surgery may be needed. Although treatment is hard work and time-consuming, it is usually successful. Very loose joints and low muscle tone mean that children need regular physiotherapy.

Eyesight

Vision problems are common. Squint (strabismus), lazy eye (amblyopia) and a drooping upper eyelid (ptosis) can occur and children will usually need to have vision checks to rule out serious visual impairment.

Heart conditions

Heart problems are routinely screened for. Holes between the chambers of the heart (septal defects) are found most frequently but most of these heal spontaneously. A small number of children have more extensive or complex heart defects that need surgery.

Mouth

A cleft palate (split in the roof of the mouth) can make feeding and later speaking difficult and usually needs surgery.

Development

The effects of a mosaic chromosome disorder are difficult to predict and what is true in general may not be true for an individual baby or child. To some extent, the effects will depend upon what proportion of cells carry the extra chromosome 9 and what proportion have the normal number of chromosomes. These proportions may differ in different parts of the body, so the proportions in blood cells may be different to those in skin cells.

Learning

Most people with trisomy 9 mosaicism will need support with their learning but a few are mildly affected and have no learning difficulties. Early intervention helps and even severely affected children may learn to read a little and write their own name.

Speech & communication

Children usually talk late. However, they are typically sociable and use a wide range of other means to make their needs and wishes felt, including signing. Some children have an unusual shape to the back of the nose and throat and this makes clear speech difficult. Regular speech therapy and access to communication devices is very helpful.

Behaviour

This is a plus point. Parents usually say that their child is calm, happy and loving. Young children who are unable to communicate are naturally frustrated but in general there are no clear patterns of difficult behaviour.

Mobility & activity

A few children reach their milestones on time, but it is more common to be late, especially for children with floppy muscles (hypotonia) and extremely mobile joints. On average babies sit around 17 months and walk with their hands held by 31 months. Walking independently for reasonable distances may have developed by the age of 5.