

Why did this happen?

A blood test to check both parents' chromosomes is needed to find out why the 10q25 or 10q26 deletion occurred. In the majority of cases the 10q25/6 deletion occurred when both parents have normal chromosomes. The term that geneticists use for this is *de novo* (dn) which means 'new'. *De novo* 10q25 and 10q26 deletions are caused by a change that occurred when the parents' sperm or egg cells formed or possibly during formation and copying of the early cells after the egg and sperm joined.

Some 10q25 and 10q26 deletions are accompanied by a gain of material from another chromosome and are often the result of a rearrangement in one parent's chromosomes. This is usually a rearrangement known as a balanced translocation in which material has swapped places between chromosomes. As no genetically important material has been lost or gained, the parent usually has no clinical or developmental problems, although they may have difficulties with fertility or childbearing. Balanced translocations involving one or more chromosomes are not rare: one person in 500 has one, making a total world population of over 13.5 million balanced translocation carriers.

Whether the deletion is inherited or *de novo*, what is certain is that as a parent there is nothing you did to cause the 10q25/6 deletion and nothing you could have done would have prevented it from occurring in your baby. No environmental, dietary, workplace or lifestyle factors are known to cause these chromosome changes. No one is to blame when this occurs and nobody is at fault.

Can it happen again?

The possibility of having another pregnancy with a 10q25/6 deletion depends on the parents' chromosomes. If both parents have normal chromosomes when their blood cells are tested, the deletion is very unlikely to happen again. However, if either parent has a chromosome rearrangement or deletion involving 10q25 or 10q26, the possibility is greatly increased of having other affected pregnancies.

Parents should have the opportunity to meet a genetic counsellor to discuss their specific recurrence risks and options for prenatal and preimplantation genetic diagnosis (PGD). PGD requires the use of in vitro fertilisation and embryo biopsy, and only healthy embryos are transferred to the mother's uterus. If the parents choose to conceive naturally, prenatal diagnosis options include chorionic villus sampling (CVS) and amniocentesis to test the baby's chromosomes. Testing is generally very accurate, although not all of these tests are available in all parts of the world.

Feeding

Most babies have a weak sucking reflex and tend to choke on liquids. Breastfeeding is rarely successful and even bottle feeding can be a struggle. However, babies can be given breast milk by bottle, cup or tube. Reflux – bringing feeds back up the food passage – is common and many newborns need to be fed by tube. Babies usually have tiny appetites and may need high-calorie milks and supplements.

Inform Network Support



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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. It was compiled by *Unique* and reviewed by Dr Veronica Mardo, John Hopkins University, USA and by Professor Maj Hultén, Professor of Medical Genetics, University of Warwick, UK. 2009 (SW)
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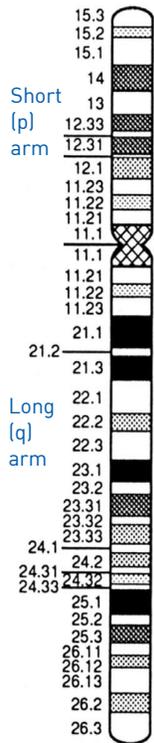
10q25 and 10q26 deletions



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What are 10q25 and 10q26 deletions?

Chromosome 10



A 10q25 or 10q26 deletion means that the cells of the body have a small but variable amount of genetic material missing from one of their 46 chromosomes – chromosome 10. For healthy development, chromosomes should contain just the right amount of genetic material (DNA) – not too much and not too little. Like most other chromosome disorders, having parts of chromosome 10 missing may increase the risk of birth defects, developmental delay and learning difficulties. However, the problems vary and depend very much on what and how much genetic material is missing.

The vast majority of 10q25 and 10q26 deletions are terminal. This means that the tip of the long arm of chromosome 10 is included in the deletion. However, some deletions are interstitial. This is where a piece of the long arm of chromosome 10 is missing, but the tip (and possibly more than just the tip) is still present.

Most children have:

- Small size and slow growth. Many babies grow slowly in the womb
- Difficulty latching on and feeding
- 'Floppy' muscles as babies, known as hypotonia. Children usually outgrow this
- Delay in skills like sitting and walking
- Some degree of learning difficulty. This is quite variable
- Squint (strabismus), with the eyes usually pointing inwards.

How common are 10q25/26 deletions?

They seem to be rare. When this leaflet was written there were around 60 people reported in the medical literature and *Unique* had 69 members with a pure 10q25/6 deletion without loss or gain of material from any other chromosome. *Unique* can put families who want contact in touch with each other.

Development

■ Motor development

All babies have some degree of developmental delay and typically sit between 8 months and 2 years. With physiotherapy and walking aids as well as support clothing, they mostly learn to walk between 18 months and 6 years. Their gait can be unsteady at first but improves with practice and maturity. However, some unsteadiness may persist so that children may need protection and supervision outdoors.

■ Learning

Most children will need some support with their learning. However, the spectrum is very broad from teenagers who have some age appropriate skills to children who have profound difficulties. Memory can be excellent and some children show particular abilities. Children with 10q25/6 deletions seem to share a love of music and singing.

■ Speech

Some delay in talking is typical and as first words usually emerge between 2 and 6 years, children can be helped by first learning to sign. While some children eventually speak conversationally, others have articulation difficulties and a minority of children continue to communicate without words.

■ Behaviour

Some older children become active and restless and find concentration difficult. Other children develop certain autistic traits, dislike touch and are wary of other children. Overall, any effects on behaviour are variable.

Medical conditions



Life is fun, and the nasogastric tube is a temporary help with feeding.

- **Growth** Most babies are small at birth and even more grow very slowly in the first year. A few catch up and go on to reach average height, but most children remain short and sometimes markedly slim.

- **Strabismus (squint)** is very common. Most squints are convergent – the eyes cross – and many children need surgery. Blocked tear ducts can occur.
- **High pain tolerance** means babies will not protest when they are in discomfort. Gastric reflux can affect them severely before there is any sign of a problem.
- **Genitals** In boys, the penis and scrotum are usually small and the hole that is usually at the end of the penis is often situated on the underside instead (hypospadias). Boys' testes have often not yet descended into the scrotum at birth.
- **Respiratory conditions** Many babies need extra oxygen as newborns and respiratory conditions remain a recurring feature of childhood. Children catch infections more often and suffer worse when they do. Older children may develop asthma.
- **Urinary tract and kidneys** Babies will have a renal scan to check their kidneys and urinary tract and parents should be told the warning signs of a urinary infection, as these are common in some children.
- **Heart conditions** affect around half of all babies. Many will clear up without treatment but some babies have complex heart conditions and need surgery.
- **Sensitive hearing** Some children have acutely sensitive hearing and are afraid of loud noises. Around one child in five has some level of hearing impairment.
- **Circulation** Some children have a low body temperature or have difficulty maintaining the temperature in their hands and feet.
- **Digestion** Chronic constipation affects almost half of *Unique* children. Dietary changes and/or medication can help to manage the problem.