

Why did this happen?

A blood test to check both parents' chromosomes is needed to find out why a 10q25/6 deletion has occurred. In the majority of cases a 10q25/6 deletion occurs when both parents have unaffected 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. Rarely, a 10q25/26 deletion is *inherited* from a similarly affected parent.

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 million balanced translocation carriers (see the Unique leaflet on [Balanced Translocations](#) for further information).

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 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 unaffected chromosomes when their blood cells are tested, the deletion is very unlikely to happen again in a subsequent pregnancy. Very rarely (less than 1% (1 in 100)), both parents have unaffected chromosomes on a blood test, but a few of their egg or sperm cells carry a chromosomal change. This is called *germline mosaicism* and it means that parents whose chromosomes appear unaffected when their blood is tested can have more than one child with the deletion.

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.

Feeding

Most babies have a weak sucking reflex and tend to choke on liquids. Breastfeeding is rarely established 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 new-borns need to be fed by tube. Babies usually have tiny appetites and may need high-calorie milks and supplements.



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

Inform Network Support



Understanding Chromosome & Gene Disorders

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www.hadds.org/ - The EBF3-HADDs Foundation was created to promote awareness, research and support for a rare genetic syndrome discovered in 2016.

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). A major revision was made by Unique (CA) in 2021/2 and reviewed by Hsiao-Tuan Chao, MD, PhD, Assistant Professor and McNair Scholar, Departments of Pediatrics-Neurology, Molecular & Human Genetics and Neuroscience, Baylor College of Medicine, US. Copyright © Unique 2015, 2022

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Understanding Chromosome & Gene Disorders

10q25 and 10q26 deletions



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What are 10q25 and 10q26 deletions?

A 10q25 or 10q26 deletion means that the cells of the body have a small 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 congenital disorders, developmental delay and intellectual disability/learning difficulties. It is important to remember that the outcome of having a 10q deletion is variable and depends on a number of factors, including what and how much genetic material is deleted.

Some 10q25 and 26 deletions are **interstitial**, 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. The majority of deletions involve 10q26 and are **terminal**, meaning that the tip of the long arm of chromosome 10 is included in the deletion. Such deletions are associated with a well-recognised condition called **10q26 deletion syndrome** (MIM #609625).

In 2009, Unique had 69 members with a pure 10q25/6 deletion without loss or gain of material from any other chromosome. By 2021, this number had reached over 200. *Unique* can put families who want contact in touch with each other.

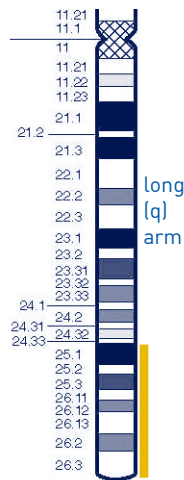
Hypotonia, Ataxia & Delayed Development Syndrome (HADDs)

In 2016, a novel neurodevelopmental syndrome named **hypotonia, ataxia and delayed developmental syndrome (HADDs)** (MIM #617330), was co-discovered by three independent research teams (Chao, Davids et al 2016, Harms et al 2016, Steven et al 2016).

HADDs is usually caused by **variants** (changes) in the nucleotide sequence of one copy of a gene called **Early B-Cell Factor 3 (EBF3)**, which is located near the tip of chromosome 10 in 10q26.3. However, deletion of only the *EBF3* gene can also cause HADDs.

The strong overlap in the features of 10q26 deletion syndrome and HADDs suggests that it is deletion of one copy of the *EBF3* gene that may be the principal cause of the features associated with distal 10q deletions, although other genes are believed to play a role in some features.

Chromosome 10



Most common features:

Every person with a 10q25/6 deletion is unique and so each person will have different medical and developmental concerns; there is known to be considerable variability, even within families with the same deletion.

- Growth delay both in the womb and after birth
- Feeding difficulties
- Hypotonia (unusually low muscle)
- Variable disabilities in learning, speech and motor development
- Problems with vision/structural eye anomalies, including strabismus (squint)
- Kidney and/or urinary tract anomalies
- Heart conditions
- Microcephaly (an unusually small head)
- Genital anomalies
- Characteristic facial features
- Seizures
- ADHD, impulsivity or autism/autistic traits

Development

■ Motor development

Children with a 10q25/6 deletion are typically slow to reach their developmental motor milestones. Most children go on to walk, climb stairs and run, although they can be unsteady with poor balance (ataxia) and co-ordination, which may put them at increased risk of falls. Many children walk with a wide gait and trip easily. Some children need support (such as a standing frame, walking frame, support boots, a supportive Lycra 'second skin' and/or leg braces) while learning to walk. Hypotonia and hypertonia can also affect fine motor skills in children with a 10q25/6 deletion.

■ 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 with first words usually emerging between 2 and 6 years. While some children eventually speak conversationally, others have articulation difficulties (apraxia of speech) and a minority of children continue to communicate without words. Any concerns around hearing should also be acted on early to help reduce any impact on speech.

■ Behaviours

While children with a 10q25/6 deletion are typically happy, sociable, loving and affectionate, a significant number of children – although not all – show a similar pattern of behavioural difficulties, including social, emotional and anxiety disorders. These include autism spectrum disorder (ASD); anxiety, attention deficit hyperactivity disorder (ADHD); sensory processing disorder (SPD); and obsessive compulsive disorder (OCD).

Medical conditions

■ **Growth** Many 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. Feeding difficulties are common.

■ **Strabismus (squint)** is very common. Most squints are convergent – the eyes cross – and many children need surgery. Other vision/structural eye anomalies can occur, including long – and short-sightedness, astigmatism and blocked tear ducts.

■ **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** Boys' testes have often not yet descended into the scrotum at birth but can be brought down by a straightforward surgical operation if they do not descend of their own accord in time. The penis and scrotum may be small. A few cases of disorders of sexual development have been reported.

■ **Respiratory conditions** Many babies need extra oxygen as new-borns and respiratory conditions can 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 are likely to 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.