Why did this happen?

To answer this question, the parents' and affected child's chromosomes need to be tested. What is certain is that, as a father or mother, there is nothing you did to cause the deletion and nothing you could have done which would have prevented it. Chromosome rearrangements affect children from all parts of the world and from all types of background. They also happen naturally in plants and animals. It is no one's fault.

The vast majority of 15q25 deletions appear to have arisen *de novo*, which means the deletion has occurred as a new event in the child, but occasionally a deletion has been inherited from a parent.

Can it happen again?

The chances of having another child with a 15q25 deletion depend on the results of chromosome tests on the parents. Where the tests show that the parents' chromosomes are unaffected, their chances of having another affected child are usually no higher than for anyone else in the population (although very rare, it can still happen due to a phenomenon known as germline mosaicism). Where the test reveals a rearrangement in the parents' chromosomes, the chances are very much higher. Each family's situation is individual, and families should be able to discuss the possibilities they face with their geneticist or genetic counsellor.

Management recommendations

- Feeding management, where necessary.
- Follow up by a developmental pediatrician.
- Physiotherapy/occupational therapy/speech and language therapy/assessment for behavioural concerns, if needed.
- Routine screening for anaemia (for deletions including proximal region of 15q25.2).
- Investigation for primary ovarian insufficiency (POI) for girls if puberty is delayed.
- Treatment for any other medical concerns e.g. possible cardiac anomalies, CDH.
- An assessment to identify the most appropriate schooling.
- A high standard of dental care is important.

Families say ...

"Take the initiative and seek help - don't be afraid to do so."

"She is a happy little girl no matter what. She is really good with her siblings and loves to play. I wouldn't change her for the world!"

Inform Network Support

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Join Unique for family links, information and support

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at www.rarechromo.org/donate Please help us to help you!

This information guide 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. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. *Unique* does its best to keep abreast of changing information and to review its published guides as needed. This information guide was compiled by Unique (CA) using information from the comprehensive *Unique* information guide for 15q25 deletions (2021).

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Understanding Chromosome & Gene Disorders

15q25 deletions

(15q25.2 microdeletion syndrome)

rarechromo.org

What is a 15q25 deletion?

A chromosome 15q25 deletion is a rare genetic condition in which there is a missing (deleted) copy of part of the genetic material that makes up one of the body's chromosomes - chromosome 15. People with a 15q25 deletion have one unaffected chromosome 15, but the other chromosome 15 has chromosomal material missing from all or part of band 15q25 on the long arm, which is divided into three sub-bands: 15q25.1, 15q25.2 and 15q25.3 (marked in red). The majority of individuals with a 15q25 deletion have a deletion involving 15q25.2, leading to the features of 15q25.2 microdeletion syndrome.

As with other chromosome disorders, having a missing piece of genetic material may increase the risk of congenital disorders (birth defects), affect the development and intellectual abilities of a child and be associated with a range of other individual features, to a varying degree. It is important to remember that the outcome of having a 15q25 deletion is variable and depends on a number of factors, including what and how much genetic material is deleted.

Features of 15q25 deletions

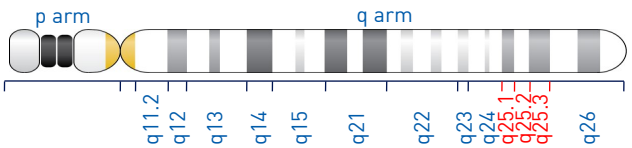
Just as "typically"-developing children can experience a number of unforeseen physical and behavioural difficulties, each person with a 15q25 deletion is unique and can have different developmental and medical concerns.
However, the most likely features associated with 15q25 deletions and/or those that are the most likely to make a difference to a child's health or development, are:

- Some degree of developmental delay (often mild to moderate)
- Some degree of intellectual disability/learning difficulty
- Short stature after birth
- Anomalies of the chest and abdomen
- Anaemia
- Thrombosis
- Heart conditions
- Cryptorchidism (undescended testes)
- Primary ovarian insufficiency (POI)
- Characteristic facial features
- Neuropsychiatric disorders (likely to be more common with deletions including the distal region of 15q25)
- Seizures (likely to be more common with deletions including the distal region of 15q25)
- Strabismus (a squint) (likely to be more common with deletions including the distal region of 15q25)

Medical concerns

- Anomalies of the chest and abdomen are one of the more common features associated with 15q25 deletions, and include reports of hernias, including congenital diaphragmatic hernia (CDH) and curvature of the spine.
- Anaemia can lead to variable symptoms, including fatigue, shortness of breath, dizziness/light-headedness and an irregular heartbeat. There is a relatively high incidence of anaemia in individuals with proximal 15q25 deletions.
- Thrombosis At least five individuals with a 15q25.2 deletion have been diagnosed with portal vein thrombosis (PVT).
- Heart conditions have been reported in association with 15q25.2 deletions. For those affected, the condition was minor and/or resolved naturally without any need for treatment or surgery.
- Anomalies of the kidneys have been found in a small number of babies.
- Primary ovarian insufficiency (POI) is characterised by the early loss of the normal function of the ovaries before the age of 40 and causes early menopause. It is thought that ~1% of women with POI have a 15q25.2 deletion.
- Anomalies of the brain are uncommon but have been reported very occasionally. A few babies are born with a head that is an unusual size or shape.
- Minor anomalies of the genitals Around half of boys with a 15q25.2 deletion had undescended testicles at birth, which can be corrected surgically if necessary.
- Hands & Feet may have minor anomalies, although there are no consistent features and not all children appear to be affected.
- Occasionally, *Unique* parents told us about minor anomalies of the eyes/problems with vision. Some children are also particularly prone to ear infections and a few children experienced some degree of hearing loss. Very rarely, anomalies of the joints, palate and dental concerns have been reported.

Chromosome 15



Development

- Growth & feeding A few babies are born small for gestation and remain small, but many babies are born within the "normal" range. While some babies and children continue to maintain a healthy growth rate, for others growth rates subsequently slow and in some cases become pronounced.

  Although some babies have no early feeding difficulties, for others feeding can be more challenging. Problems are often temporary, but in a few cases we know that difficulties led to poor weight gain and failure to thrive. Longer term, a few *Unique* children had a very limited diet and some may have had sensory issues around food.
- Gross & fine motor skills There appears to be considerable variability in the extent to which gross motor skills are affected. Some reach milestones such as rolling, sitting and walking around the same age as "typical" children; others experience some degree of delay, usually mild to moderate, which may show a marked improvement with time. Some babies and children have reduced muscle tone (hypotonia), although muscle tone often improves with age. Some degree of difficulty with hand use and hand-eye coordination appears to be common.
- Speech Speech is typically one of the most commonly affected areas of development. Difficulties with articulation, which make it difficult to make clearly intelligible speech sounds and can make communication with strangers a challenge, were referred to by many *Unique* parents and often persisted. With time, it appears that older children tended to use long, complex sentences.
- Learning There is considerable variability in learning ability. For some, learning appears to be unaffected; for others some degree of learning difficulty or intellectual disability, usually mild to moderate, has been recorded.
- Social, emotional & anxiety disorders *Unique* families told us about children who are kind, thoughtful, funny, caring individuals. Children may find social interactions challenging and they may be somewhat anxious and shy around strangers or in new social situations.

  The 15q25.2 region has been linked to an increased risk of developing neurodevelopmental and neuropsychiatric disorders. These include social, emotional and anxiety disorders, such as autism spectrum disorder (ASD); anxiety, attention deficit hyperactivity disorder (ADHD); sensory processing disorder (SPD); and seizures. There is some suggestion that individuals with deletions involving the more distal region may be more susceptible to these than those involving the proximal region alone.