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 15q14 deletions appear to have arisen *de novo*, which means the deletion has occurred as a new event in the child, but occasionally a deletion had been inherited from a parent.

Can it happen again?

The chances of having another child with a 15q14 deletion depend on the results of chromosome tests on the parents. Where the tests show that the parents' chromosomes are normal, 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 all 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.
- Referral to a specialist cleft team, if needed.
- Cardiologic follow-up where an individual has a 15q14 deletion including the *ACTC1* gene, even when no heart defect is present at birth.
- An assessment to identify the most appropriate schooling.
- Dental problems are very common in children with chromosome disorders and a high standard of dental care is important.

Families say ...

"Each child is different. You have to look at their needs as an individual. We have found time is the best indicator of what our son's needs are and we have come to understand him much better over the years."

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!

Version 1 (CA)

Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
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Understanding Chromosome & Gene Disorders

15q14 deletions

rarechromo.org

What is a 15q14 deletion?

A chromosome 15q14 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 15q14 deletion have one unaffected chromosome 15, but the other chromosome 15 has chromosomal material missing from all or part of band 15q14 on the long arm (marked in red). Some people also have larger deletions that extend into other bands e.g. 15q12, 15q13, 15q15.

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 15q14 deletion is variable and depends on a number of factors, including what and how much genetic material is deleted.

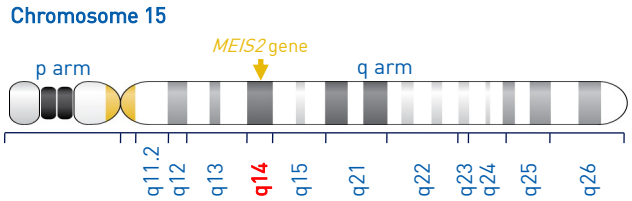
Features of 15q14 deletions

Just as "typically"-developing children can experience a number of unforeseen physical and behavioural difficulties, each person with a 15q14 deletion is unique and can have different developmental and medical concerns. However, the most likely features associated with 15q14 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
- Some degree of intellectual disability
- Speech delay/difficulties with speech
- Anomalies of the palate e.g. cleft palate, bifid uvula, a high/arched palate
- Low muscle tone (hypotonia)
- Joint hypermobility (laxity)
- Feeding difficulties
- Heart anomalies
- A small head (microcephaly)
- Short stature
- Undescended testes at birth in boys (cryptorchidism)
- Café-au-lait spots (CALs)
- Social, emotional and anxiety disorders
- Frequent ear infections/glue ear, which usually resolve during childhood
- Minor anomalies of the hands and feet
- Minor dental concerns
- Characteristic facial features

Medical concerns

- **Anomalies of the palate** Anomalies of the palate (roof of the mouth) have been observed in the majority of babies and children, and can cause difficulties with feeding, hearing, teething and speech production.
- **Heart** A heart condition has been reported for just under half of those with a 15q14 deletion in medical literature, including deletions extending to neighbouring bands. Although heart conditions were associated with some deletions (and mutations) in the *MEIS2* gene, they appear to be more common where the deletion also includes the *ACTC1* gene.
- **Head** An unusually small head (microcephaly) is relatively common.
- **Limbs & joints** Joint hypermobility appears to be relatively common and may affect fine and gross motor skills. It is sometimes associated with pain and stiffness in the joints and muscles and joints that dislocate easily.
- **Minor anomalies of the genitals (boys)** Some boys may be born with undescended testis/testes (cryptorchidism).
- **Café-au-lait spots (CALs)** CALs have been reported in a number of individuals with deletions that include the *SPRED1* gene.
- **Hands & Feet** Children may have minor anomalies of the hands and feet, although there are no consistent features and not all children appear to be affected.



The *MEIS2* gene

Location:15q14 [37,183,222 - 37,393,500 (GRCh37/hg19)
15q14 [36,889,204 - 37,101,311 (GRCh38/hg38)

The *MEIS2* (Meis Homeobox 2) gene codes for a protein that helps control the expression of other genes and has been shown to play a role in controlling the growth of the developing embryo during pregnancy. It has been suggested that having a missing copy of *MEIS2*, or other mechanisms that lead to under-expression of the gene, may contribute to the features associated with 15q14 deletions, including anomalies of the palate, characteristic facial features, congenital heart defects and autism spectrum disorder (ASD).

Development

- **Growth & feeding** For many, growth patterns appear to be within the normal range, but some babies and children have a short stature and/or an unusually small head.

  Although some parents told us that their baby had no early feeding difficulties, for others feeding was more challenging. Anomalies of the palate may exacerbate these problems. Some babies experienced reflux, colic or constipation. Problems generally appear to have been temporary and didn't persist into childhood.
- **Gross & fine motor skills** Babies and children are likely to take more time to reach their developmental milestones, including rolling, sitting and walking. This delay was often mild to moderate and most had learned to walk somewhere between the ages of 14 months and three years. Some had reduced muscle tone (hypotonia) or occasionally increased muscle tone (hypertonia). Cases of hypermobile joints and developmental co-ordination disorder (DCD) (dyspraxia) were also reported. It seems that difficulties with hand use and hand-eye coordination are to be expected.
- **Speech** Information suggests that some degree of language delay or difficulty with speech appears to be common, although there is considerable variation and some children appear to be unaffected. It has been suggested that genes within 15q14 may be associated with oral-motor skills, articulation and phonological memory. Anomalies of the palate can also affect speech. Limited information from *Unique* parents indicated that there was improvement with time and several children used long, complex sentences.
- **Learning** Some degree of intellectual disability, often mild to moderate, seems to be a common feature of 15q14 deletions. It has been suggested that 15q14 deletions where the *MEIS2* gene is completely deleted, or that involve other genes or extend into other bands, may be more likely to be associated with a moderate to severe ID.
- **Social, emotional & anxiety disorders** The testimony we have speaks to children who are fun, loving individuals. Some children have received a diagnosis for a specific social, emotional or anxiety disorder, including an autism spectrum disorder (ASD), anxiety, attention deficit hyperactivity disorder (ADHD) and sensory processing disorder (SPD), although they don't appear to be a consistent feature.

This information guide was compiled using information from the comprehensive *Unique* information guide for 15q14 deletions (2020). Please refer to the longer guide for further details.