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 duplication 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.

5q14 duplications are known to be either inherited from a parent or to occur *de novo* (dn), which means the duplication has occurred as a new event in the child.

Can it happen again?

The chances of having another child with a 5q14 duplication 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 if necessary e.g. attending a feeding clinic to help with eating, drinking and swallowing difficulties.
- Follow up by a developmental pediatrician.
- Physiotherapy/occupational therapy/speech and language therapy/assessment for behavioural concerns if needed.
- Eye examination e.g. check if patching, exercises and/or glasses are needed.
- 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 ...

"We are very involved at school and at home to ensure all the tools, therapies, etc. are provided. We also try many different activities e.g. sports and music, to find what our son enjoys and excels at. Do not give up hope, we have seen incredible progress in our son."

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!

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Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
Company Number 5460413

Understanding Chromosome & Gene Disorders

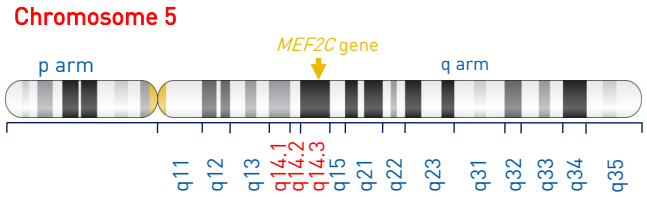
5q14 duplications

rarechromo.org

What is a 5q14 duplication?

A chromosome 5q14 duplication is a rare genetic condition in which there is an extra copy of part of the genetic material that makes up one of the body's 46 chromosomes - chromosome 5. A duplication is also called a partial trisomy.

People with a 5q14 duplication have one normal chromosome 5, but the other chromosome 5 has an extra piece of chromosomal material from all or part of band q14 on the long arm, which includes q14.1, q14.2 and q14.3.



As with other chromosome disorders, having an extra piece of genetic material may increase the risk of 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 5q14 duplication is variable and depends on a number of factors, including what and how much genetic material is duplicated.

Common features

Just as "typically"-developing children can experience a number of unforeseen physical and behavioural difficulties, each person with a 5q14 duplication is unique and can have different developmental and medical concerns. However, the most likely features associated with 5q14 duplications 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)
- Some degree of intellectual/learning disability
- Delayed/absent speech
- A small head (microcephaly) (5q14.3 duplications including *MEF2C*)
- Autism or autistic traits (5q14.1 duplications)

Other features have been noted in association with a 5q14 duplication. Some are known to be generally more common in children with chromosome disorders; others may in fact be unconnected with the chromosome disorder.

Medical concerns

- **General well being** Although the majority of *Unique* families describe their child's general state of health as "good" or "very healthy", a few children have on-going health conditions, including recurrent chest infections, asthma and seizures.
- **Head shape** A small head size (microcephaly) is one of the most consistent features of 5q14.3 duplications and has been reported in medical literature to be associated with upregulation of the *MEF2C* gene.
- **Eyes & vision** Problems with vision, both short- or long-sight, have been reported for several children, and a few had a squint (strabismus).
- **Seizures** Seizures, including epilepsy, are not thought to be a common feature of 5q14 duplications but have affected some people.
- **Brain anomalies** Under MRI, a number anomalies of the brain have been reported, but many were mild and many individuals appear to be unaffected.
- **Heart** The *MEF2C* gene plays a role in heart development but reports of heart conditions associated with 5q14 duplications are extremely rare.
- **Hands & feet** Children with a 5q14 duplication often have minor anomalies of the hands and feet.
- **Hearing** Frequent ear infections appear to be common. These can sometimes lead to a build-up of fluid in the middle ear, called glue ear. Glue ear usually resolves as children get older.
- **Eczema** Several children with a 5q14.3 duplication including *MEF2C* have eczema, where the skin becomes red, itchy and inflamed.

The *MEF2C* gene

Location: 5q14.3 [88,014,058 - 88,199,922 (GRCh37/hg19)]
During development of the human body, the *MEF2C* (Myocyte Enhancer Factor 2C) gene interacts with other genes in a multigene network, playing a crucial role in brain development, as well as the development of the heart, blood vessels, immune system, muscles and face. The presence of an extra copy of *MEF2C*, is one of the mechanisms that lead to overexpression of the related protein. This may affect this carefully controlled network of genes, for instance by causing the upregulation and overexpression of other genes in the network. This is thought to lie behind many of the features associated with 5q14 duplications.

Development

- **Growth & feeding** Some babies we know about had no early feeding difficulties; for others feeding was more challenging. Problems were often temporary, but in a few cases difficulties led to poor weight gain and, very rarely, failure to thrive. The few children we know about with larger 5q duplications including part of 5q14 appear to have experienced more problems with feeding.
  Longer term, some children with a 5q14.3 duplication including *MEF2C* seem to be on the small side; others are nearer average weight and height. There is limited evidence to suggest that 5q14.1 duplications (not including *MEF2C*) are associated with overgrowth or being overweight.
- **Gross & fine motor skills** It appears that many babies and children are late to achieve their gross motor skill 'milestones' but the delay is often mild. It generally takes longer than is typical for babies and children to roll over, sit, crawl and walk, although many children start to walk independently between 18 months and two years. Difficulties with hand use and hand-eye coordination appear to be common among *Unique* children.
- **Speech** The limited information we have suggests that speech is typically one of the most commonly affected areas of development. Speech and language development is often severely delayed, regardless of duplication size, and a few children remain non-verbal; others go on to develop a good level of speech. Parents often told us that that their child's comprehension of language is better than their ability to communicate using language.
- **Learning** It appears that children are very likely to need early and ongoing support with their learning. Some children we know about have mild to moderate learning disability, while others are more severely affected.
- **Behaviour** The testimony we have speaks to children who are happy, loving, funny and caring. Some parents told us that their child's mood and behaviour could suddenly change, leading to challenging behaviours. Many parents told us that their child has experienced issues around sleep, although these often appear to improve with age.
- **Social, emotional & anxiety disorders.** Some children, almost exclusively with a 5q14.1 duplication, have received a diagnosis for a specific social, emotional or anxiety disorder, including an autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD).

See long guide for more details relating to all these features.