How common are15q11q13 duplications?

There is no official estimate of the prevalence of interstitial 15q11q13 duplications. Many children will not have been diagnosed and not all diagnosed children are reported in the literature or have joined a support group. Currently there are less than 100 people reported in the medical literature (Isles 2016).

Why did this happen?

When sperm and egg cells are formed, or after they join to form a single cell from which a child is formed, copies of all genetic material must be made. The biological copying method is not perfect and occasionally duplications and other variations occur.

It is important to know that as a parent there is nothing you could have done to prevent this from happening. No environmental, dietary or lifestyle factors are known to cause interstitial 15q11q13 duplications. There is nothing that either parent did before or during pregnancy that caused the duplication.

Can it happen again?

If a parent has the same duplication as their child, the possibility of having another child with this duplication is 50% in each pregnancy. If neither parent is found to have this chromosomal change, it is unlikely that they will have another child with a 15q11q13 duplication.However, very rarely, parents are identified as having unaffected chromosomes by a blood test, but a few of their egg or sperm cells carry a chromosomal change. This is called germline mosaicism and means that such parents can have more than one child with the same chromosome disorder. If they wish, parents can discuss with a genetic counsellor the specific recurrence risks.

Families say ...

" Join a support group. It gets better and they enrich your life in unexpected ways. There is always some grief, but you learn to celebrate different milestones. "

" Just love your children and do your best to help them to improve, they are normal children and want to be loved like the others. Let them have more autonomy and freedom if you can, it will help them. "

" Get one doctor (e.g. paediatrician) to be main point of contact. Our paediatrician only knew about dup15 by googling. We did all of the research, made appointments etc. ourselves. At one stage we had 15 specialists. It would have been nice to have one contact to guide us and make suggestions. "

" At school, make sure you and the support network are on the same page. Some teachers don't 'get it'. "

" Get your child a good psychologist - your child needs to vent and to get guidance on friendships etc. "

Inform Network Support

Rare Chromosome Disorder Support Group,
The Stables, Station Road West,
Oxted, Surrey. RH8 9EE. UK.
Tel +44(0)1883 723356
info@rarechromo.org
www.rarechromo.org

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 (AP)

Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
Company Number 5460413

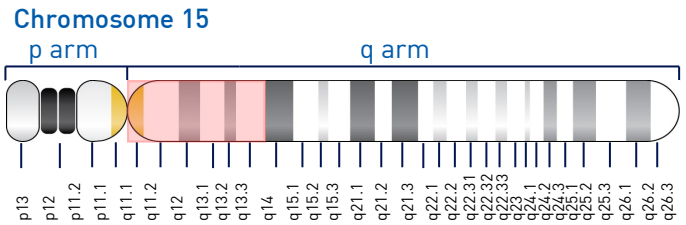
Understanding Chromosome & Gene Disorders

15q11q13 duplications (interstitial)

rarechromo.org

What is a 15q11q13 duplication (interstitial)?

A 15q11q13 duplication is a rare genetic condition caused by a duplicated piece of genetic material from one of the body's 46 chromosomes – chromosome 15. An interstitial duplication, means that the duplicated piece of DNA is added to an existing chromosome (usually next to the original piece of chromosome). q11q13 describes the location of the piece of genetic material that has been duplicated as shown in the image below (shaded in pink), it is located on the q arm of chromosome 15 within bands 11 to 13.



How will this affect me or my child?

For typical and healthy development, chromosomes should contain the expected amount of genetic material. Having an extra piece of chromosome 15 can affect a child's development and intellectual abilities.

The features associated with 15q11q13 duplications are very variable and depend on a number of factors including what and how much genetic material is duplicated and whether the duplication is from the chromosome 15 passed on by the mother or by the father. This is because this region of chromosome 15 contains imprinted genes. These are genes that are switched off in either the mother's chromosome 15 or that of the father. Gene imprinting is a natural process, presumed to regulate appropriate gene activity.

This guide is a summary of important findings relating to interstitial 15q11q13 duplications. For more detailed information relating to this duplication, please read *Unique*'s full length information guide for 15q11q13 duplications (interstitial).

Possible Features

It is important to note that some people do not appear to be affected by their 15q11q13duplication, while others may have one or a number of features. The following is a list of some of the possible features:

- Learning difficulties or intellectual disability
- Speech and language difficulties or absent speech
- Autism spectrum disorder or other behavioural difficulties
- Seizures including infantile spasms
- Developmental delay
- Sensory processing disorder
- Anxiety and/or emotional lability (rapid change in type/strength of emotion)
- Hypotonia (low muscle tone)
- Smaller or larger head size
- Slightly unusual facial features

Medical concerns

The most commonly reported medical causes of concern for children with an interstitial 15q11q13 duplication are seizures and gastrointestinal problems. However, most families report that their child is consistently in generally good health.

Seizures

About half of the families known to *Unique*, reported their child has or has had seizures (a sudden change in electrical activity of the brain that causes momentary brain dysfunction), the other half reported that their child had no signs of any seizure activity (with ages ranging from baby to adulthood). The age at which seizures start varies between baby and adulthood and often only a few seizures occur, or seizures stop after a certain amount of time.

Gastrointestinal problems

Approximately 3/4 of families known to *Unique* reported their child with an interstitial 15q11q13 duplication had some form of gastrointestinal problem. Constipation was commonly reported as was reflux [gastroesophageal reflux (GERD) this is when food returns up the food pipe].

Development

Developmental delay has been described by over half of *Unique* families who shared this information about their child, ranging from mild to severe/global.

Behaviour Diagnoses that have been associated with people with a 15q11q13 duplication are as follows:

- Autism or ASD: Autism spectrum disorder
- ADHD: Attention deficit hyperactivity disorder
- OCD: Obsessive compulsive disorder
- SPD: Sensory processing disorder
- PDD-NOS: Pervasive developmental disorder (now known as ASD)
- Anxiety

Feeding and Growth About half of families known to *Unique* mentioned that their child with an interstitial 15q11q13 duplication had feeding problems as a baby.

Sleep Almost half of families known to *Unique* remarked that their child has (or has had) some form of regular sleep disruption.

Motor skills and Mobility Roughly half of families known to *Unique* mentioned that their child's gross motor skills are affected. This is likely to be related to hypotonia. The same number of families also mentioned their child with an interstitial 15q11q13 duplication has difficulties with fine motor skills ranging from mild to severe.

Ability to learn Most children described in the literature and members of *Unique* experience learning difficulties. While some children are identified as having a mild learning difficulty, the learning abilities of less than half reported are more severely affected and they are diagnosed as having intellectual disability.

Speech, communication and sociability Most families known to *Unique* mentioned their child with an interstitial 15q11q13 duplication had or has delayed speech. Parents note that their children's comprehension of language is better than their ability to communicate using language. While children do develop language skills at varying levels, a number of families mentioned their child did not start to speak until age 4 or 5 years. Non-verbal children (over the age of 5) with an interstitial 15q11q13 duplication have been reported.