Why did this happen?

It is important that you know the deletion was not caused by anything you did. When children are conceived, their parents' genetic material is copied in the egg and sperm that makes that child. The biological copying method is not perfect and random, rare changes occur. Each person has their own unique set of genetic changes, that they would not normally know about unless an important gene, or number of genes were affected. For some children, the deletion has been 'inherited' from an unaffected parent.

Could this microdeletion affect my other children?

A 10q11.22q11.23 deletion may have been a 'brand new' change in your child (this is known as a *de novo* change), so will not be found in either parent. If parental DNA, from a blood sample, is tested and the deletion is not found, it is very rare but still possible for the deletion to be inherited by a future child. This is due to a phenomenon known as germline mosaicism, when the deletion is present in a few egg and sperm cells. The chances of this happening are less than 1%.

If the deletion is 'inherited' from a parent, there is a 1 in 2 (50%) chance of a child's siblings inheriting the same deletion.

Is there more information or future research?

So far, research has found some potential genes within the 10q11.22q11.23 region that, when deleted or changed, may affect a child's development, but the reason why children with a 10q11.22q11.23 deletion experience difficulties is not fully understood.

Very rarely, a missing copy of one gene in the deletion, and a genetic change (variant/mutation) in the other copy of the gene on the other chromosome 10 has been identified. Further research is needed to find out more about these potential conditions.

If your child has extra unexpected or severe clinical features then they may benefit from being referred to your local Clinical Genetics Team for further assessment.

Families say ...

" There does not seem to be any information on this deletion. Doctors don't seem to be very concerned either. I find it isolating having a son with this deletion as not a lot is known about it. " - Age 8

" I attended autism training workshops. I spoke to an Educational Psychologist about his PDA. Trial and error. Have tried it all and have learnt to work around his needs and sensitivity. Usual autism strategies such as reward charts etc. have not worked as he feels pressured. It is a matter of picking your battles with him and working around his anxiety. When things get too much, I reach out for help. " Age - 8

Inform Network Support

Rare Chromosome Disorder Support Group,
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Tel: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

Facebook groups
www.facebook.com/groups/chromosome10disorder/
www.facebook.com/groups/152331614838414/

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 leaflet was written by Dr Corrina Powell, Specialist Registrar in Clinical Genetics, Rosa SpencerTansley, Trainee Genetic Counsellor, Leicester Royal Infirmary Clinical Genetics Department ,UK and *Unique* (AP).

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Rare Chromosome Disorder Support Group
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Understanding Chromosome & Gene Disorders

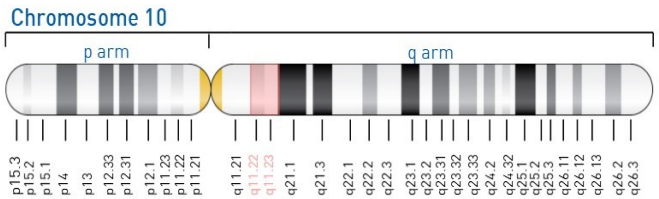
10q11.22q11.23 deletions and microdeletions

rarechromo.org

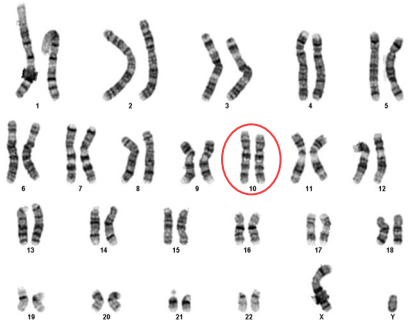
This leaflet aims to briefly summarise what is known about 10q11.2q11.23 deletions/microdeletions and how having this deletion may affect you and/or your child. More information is available in the full *Unique* information guide. You can also speak with your Doctor and/or Genetic Counsellor. By joining *Unique* you will be able to contact other families who have the same or similar deletion.

What is a 10q11.2q11.23 deletion?

A deletion or microdeletion means there is a 'small' amount of genetic material missing, in this case, from chromosome 10. The deletion is on the long arm (q) of chromosome 10 and spans two regions known as bands 11.22 and 11.23 (shown in pink in the image below). A 'detailed chromosome test' called a microarray, will have found this deletion or microdeletion.

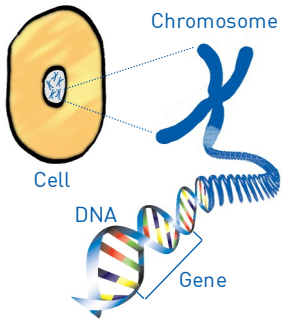


Most of our cells have 23 pairs of chromosomes; one chromosome from each pair is usually inherited from each parent. Unless you've been informed otherwise, your child (and possibly yourself or your partner) has one chromosome with this deletion, the other copy of chromosome 10 is normally unaffected.

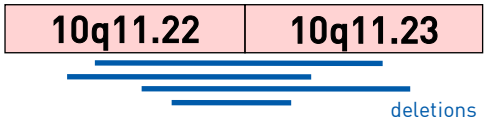


Chromosomes pairs 1-22, X and Y (male)
Chromosome pair 10 is circled in red

Chromosomes are made from DNA and DNA codes for our genes. Our genes act like 'instructions' that tell our body how to grow, develop and function.



Region 10q11.22q11.23 contains a large amount of DNA that codes for over 100 genes. Not all 10q11.22q11.23 deletions are the same, but most will share a common region. Larger deletions include more genes. Some example deletions are shown as blue lines in the image below.



What are the symptoms?

Each person with a 10q11.22q11.23 deletion may be affected differently and some people do not have any symptoms. Even siblings with the same parents and the same deletion can have different symptoms or features. There is currently some evidence in the medical journals and databases to suggest this deletion may be associated with developmental delay and learning difficulties. Speech and language difficulties have also been identified in a few children with this deletion as well as behavioural difficulties such as autism or autistic like features and ADHD or similar features.

- Developmental delay / learning difficulties (particularly speech delay)
- Behavioural difficulties [e.g. autism spectrum disorder (ASD), attention deficit disorder (ADHD) or ASD/ADHD like features, anxiety]

Ability to learn

Some, but not all children affected by their 10q11.22q11.23 deletion experience learning difficulties. Some children are identified as having mild to moderate difficulties but others are more severely affected and have been given a diagnosis of intellectual disability (ID).

Speech and language

Some *Unique* families with a child with a 10q11.22q11.23 deletion informed us that their child had or has delayed speech. An assessment by a speech and language therapist may be helpful.

Behaviour

Diagnoses that have been reported in some children with a 10q11.22q11.23 deletion include ASD (autism spectrum disorder) and ADHD (attention deficit hyperactivity disorder) but only a few children have been given these diagnoses. Parents have also mentioned their children experienced anxiety.

Medical concerns

At the moment (2020) there doesn't appear to be any reason for concern regarding the overall health of children with a 10q11.22q11.23 deletion. There is not currently much information available but *Unique* families have mentioned their child with this deletion is generally in good health.

Seizures

A few children with a 10q11.22q11.23 deletion have been reported in the medical literature as having seizures. Only one *Unique* member is known to have had seizures (absence seizures at a young age that resolved with a course of medication).

Feeding

A few babies with a 10q11.22q11.23 deletion are known to have experienced feeding problems such as reflux (when food returns up the food pipe from the stomach).

Motor skills

Some children with a 10q11.22q11.23 deletion have been reported as starting to walk late and continued to show some difficulties in early childhood. For some, this may be related to hypotonia and/or joint hypermobility. Some children have also been described as having mild difficulties with fine motor skills such as poor hand use and coordination.