How common are 2q13 microdeletions?

It is difficult to estimate the prevalence of 2q13 microdeletions, however there are about 30 case reports in the medical literature to date (2018) and over 150 people with one of these deletions have been noted in 'copy number variant' databases. *Unique* currently has 44 members (32 families) worldwide with a deletion that includes region 2q13, 29 of these members (19 families) do not have any known addition genetic diagnoses.

Why did this happen?

When a sperm and egg cell join they form a single cell and this cell must continuously make copies of itself (and all its genetic material) in order to produce the billions of cells that are necessary for human growth and development. Sometimes during this process, parts of a chromosome are lost, duplicated and/or become rearranged.

In about half of the children identified so far, their 2q13 microdeletion was inherited from a parent. The other half have a de novo (dn) deletion which means the deletion occurred as a new event in that child.

It is important to know that as a parent there is nothing you could have done to prevent the deletion from happening. No environmental, dietary or lifestyle factors are known to cause 2q13 microdeletions. There is nothing that either parent did before or during pregnancy that caused the microdeletion.

Can it happen again?

Where both parents are thought to have unaffected chromosomes, it is unlikely they will have another child with a 2q13 microdeletion (the chance is less than 1%).

In families where the 2q13 microdeletion has been inherited from a parent, the possibility of having another child, either a girl or a boy, with the 2q13 microdeletion rises to 50% in each pregnancy. However, the effect of the microdeletion on the child's development, health and behaviour cannot be predicted.

Families say ...

> " He is a lovely, cuddly affectionate boy who gets frustrated by his own limitations. Take each day as it comes, don't try to predict what will happen. "

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!

Websites, Facebook groups and other links:

Unique has a number of public and private facebook groups worldwide
https://www.facebook.com/groups/chromo2syndromes/ is a chromosome 2 disorder support group

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 booklet was compiled by Lucy Williams, NHS, UK and *Unique* (AP) using information from the comprehensive *Unique* information guide for 2q13 microdeletions written in 2018.

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Rare Chromosome Disorder Support Group
Registered in England and Wales

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Understanding Chromosome & Gene Disorders

2q13 Microdeletions

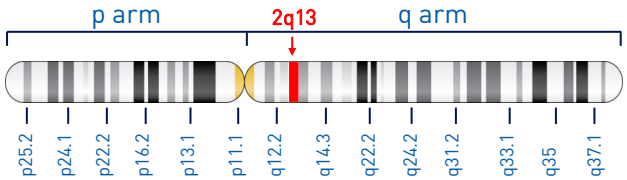
rarechromo.org

What is a 2q13 microdeletion?

A 2q13 microdeletion is a rare genetic condition caused by a small piece of missing genetic material from one of the body's chromosomes - chromosome 2.

Having a missing piece of chromosome 2 may affect the development and intellectual abilities of a child. The outcome is very variable and depends on a number of factors including what and how much genetic material is missing.

Chromosome 2



Each chromosome has a short (p) arm and a long (q) arm. Bands are numbered outwards starting from where the short and long arms meet, at a point called the centromere (coloured yellow in the image above). Region 2q13 is on the q arm of chromosome 2 in band 13 close to the centromere (highlighted in red and indicated with a red arrow in the image above).

How will this affect me or my child?

For typical and healthy development, chromosomes should contain the expected amount of genetic material. Having a missing piece of chromosome 2 may affect a persons development and intellectual abilities.

A deletion of genetic material can occur anywhere within band 2q13 but two different and recurrent deletions have been identified in a number of people. Both of these deletions have been reported in children with developmental delay and learning and/or behavioral difficulties. These deletions have also been found in the 'general population' and parents with mild or no apparent difficulties (one more so than the other).

This guide is a summary of important findings relating to 2q13 microdeletions. For more detailed information relating to these deletions, please read *Unique*'s full length information guide for 2q13 microdeletions.

Possible Features

Features of any genetic change can vary considerably but some effects of having a 2q13 microdeletion appear to be more common than others. The following is a list of possible features:

- Low muscle tone (hypotonia)
- Developmental delay
- Learning difficulties or intellectual disability
- Smaller or larger head size
- Speech and language difficulties
- Autism spectrum disorder or other behavioural difficulties
- Anxiety
- Seizures

Medical concerns

Seizures

Reports in the medical literature have noted that some children with 2q13 microdeletions (4 out of 22) have seizures. A few *Unique* families have also mentioned their child has experienced seizures.

Heart

A congenital heart defect has been identified in a few children reported in the medical literature and known to *Unique*.

Ears and Hearing

Several *Unique* members have mentioned that their child with a 2q13 microdeletion suffered numerous ear infections when young.

Eyes and Vision

Although eye and vision problems are not commonly reported, 7 *Unique* families have reported that their child has an eye or vision problem.

Brain and Head Size

A small head and brain (microcephaly) has been found in 6/22 children reported in the medical literature with a 2q13 microdeletion and two children were identified as having a large head and brain (macrocephaly).

Development

Physical Development

Since features associated with 2q13 microdeletions are so variable, babies and children are diagnosed at various stages of development. Those who are born with obvious physical problems are more likely to be tested at birth. Others may be offered a genetic test if other unusual physical features are observed. Some children are identified as babies or toddlers due to concerns such as developmental delay, floppiness, feeding problems or lack of eye contact. Others are identified due to behavioural characteristics or learning difficulties.

Learning

Most children with a 2q13 microdeletion described in the literature and members of *Unique* experience learning difficulties. Some children are identified as having a mild learning difficulty, others are more severely affected, and they are diagnosed as having intellectual disability.

Behaviour

Not all children or adults with a 2q13 microdeletion have behavioural difficulties, but as a group, they appear to show a higher incidence of behavioural, social and communication difficulties.

Speech and language

A number of children with a 2q13 microdeletion are known to have or have had delayed speech, word retrieval problems, difficulty understanding abstract concepts, long and short term memory problems or an auditory processing disorder (the way the brain processes sounds).

Feeding

Some *Unique* families have mentioned feeding problems either as a baby or a child/adolescent or both.

Sleep

It is common for *Unique* families who have a child with a 2q13 microdeletion to mention that their child has some form of sleep disruption. Children can find it difficult to 'switch off' and fall asleep at night, some children do not sleep for long periods of time and wake repeatedly in the night, some wake far too early in the morning.