How common are 16p11.2 microduplications?

It is difficult to estimate the true prevalence of 16p11.2 microduplications since not everyone has had a genetic test. However recent studies have found that it may occur in roughly 1 person in every 3000. It is slightly more common in people with language and or mental health difficulties. *Unique* currently has over 200 members with a 16p11.2 microduplication (2020).

Why did this happen?

When children are conceived their parents' genetic material is copied into the egg and sperm that makes a new child. During this complicated process rare changes occur in the chromosomes of children that are not seen in the DNA of their parents. This is known as *de novo* (dn), which means the genetic change occurred as a new event in that person.

It is important to know that as a parent there is nothing you could have done to prevent the duplication from happening. No environmental, dietary or lifestyle factors are known to cause 16p11.2 microduplications. There is nothing that either parent did before or during pregnancy that caused the change. We all have genetic changes in our chromosomes but it's only when important genes are affected that we notice an effect.

16p11.2 microduplications can also be inherited from an unaffected or affected parent.

Can it happen again?

Where both parents are thought to have unaffected chromosomes, it is unlikely they will have another child with a 16p11.2 microduplication [there is still a small chance (<1%) due to a phenomenon known as germline mosaicism, where egg or sperm cells carry the change but other cells of the body do not].

In families where the 16p11.2 microduplication has been inherited from a parent, the possibility of having another child, either a girl or a boy, with the duplication is 50% in each pregnancy. However, the effect of the microduplication on a child's development, health and behaviour cannot be predicted.

Families say ...

"She is a joy to be around. She has taught us so much. The way she sees the world with no worries and no fear is inspiring!"

"As a child, he had the most beautiful, honest nature about him. Although he had mental setbacks, he had such a sweet nature with no comprehension or ability to be mean."

"He has a dry sense of humour, loving in his own little way, wouldn't change him one bit. His bravery and courage is amazing."

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!

Facebook groups:

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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 *Unique* (AP) using information from the comprehensive *Unique* information guide for 16p11.2 microduplications updated in 2020.

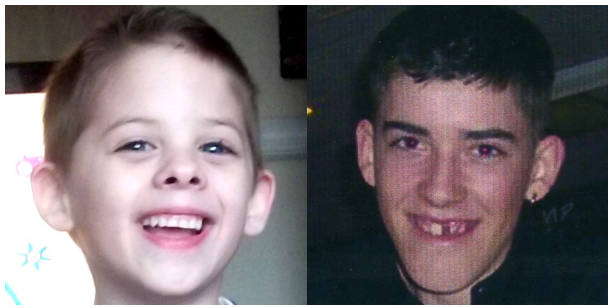
Version 1 (AP)

Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
Company Number 5460413

Understanding Chromosome & Gene Disorders

16p11.2 Microduplications



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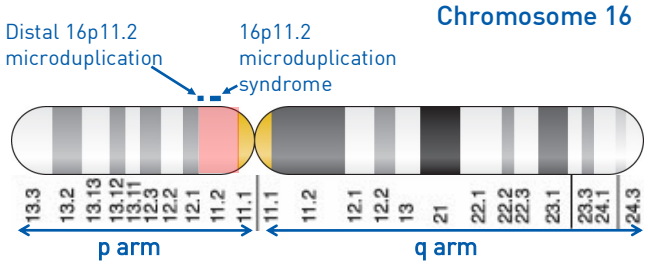
What is a 16p11.2 microduplication?

A 16p11.2 microduplication is a genetic condition caused by a small piece of extra genetic material from one of the body's chromosomes - chromosome 16.

Any piece of chromosome 16p11.2 can be duplicated but two different and recurrent microduplications have been identified in a number of people (as shown in the image below). Both of these duplications have been reported in children with developmental delay and learning, speech and/or behavioral difficulties. These duplications have also been found in the 'general population' and parents with mild or no difficulties.



Region 16p11.2 is on the p arm of chromosome 16 in a region known as 11.2 (shaded red 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 an extra piece of chromosome 16 may affect the development, behaviour and intellectual abilities of a child or adult. The symptoms are variable and depend on a number of factors including what and how much genetic material is duplicated, the unique genetic background of each person and environmental factors.

This guide is a summary of important findings relating to the two most common microduplications of 16p11.2. For more detailed information relating to these duplications, and differences between them, please read *Unique*'s full length information guide for 16p11.2 microduplications.

Possible Features

Features of any genetic change can vary considerably but some effects of having a 16p11.2 microduplication appear to be more common than others. The following is a list of possible features:

- Delays in speech and language development
- Learning difficulties or intellectual disability
- Some delay in learning to sit, move and walk
- Increased likelihood of difficult behaviour
- Increased susceptibility to neurodevelopmental disorders including ADHD (attention deficit hyperactivity disorder) and ASD (autism spectrum disorder)
- Increased susceptibility to mental health difficulties
- Possible tendency to be underweight (for 16p11.2 microduplication syndrome)
- Possible growth delay or short stature (for distal 16p11.2 microduplications)
- Possible vulnerability to seizures

Medical concerns

Seizures

Reports in the medical literature have noted that some children with a 16p11.2 microduplication have seizures. Some *Unique* families have also mentioned their child has experienced seizures.

Heart

A congenital heart defect has been identified in a few children with a 16p11.2 microduplication.

Head and Brain Size

A small head and brain (microcephaly) has been found in some children with a 16p11.2 microduplication.

Curvature of the spine

Some children and adults with a 16p11.2 microduplication have been diagnosed with scoliosis.

Low muscle tone (hypotonia)

It is not uncommon for babies with a 16p11.2 microduplication to have low muscle tone (hypotonia).

Development

Since features associated with 16p11.2 microduplications are so variable, babies and children are diagnosed at different stages of development. Those who are born with obvious physical problems, such as a heart defect, are more likely to be offered a genetic test at birth. Babies or toddlers are likely to be offered a test due to concerns such as developmental delay, floppiness (hypotonia), feeding problems or lack of eye contact. Older children and adults may be identified due to behavioural characteristics or learning difficulties.

Learning

Most children affected by their 16p11.2 microduplication experience learning difficulties. Some children are identified as having mild learning difficulties that can improve over time while others are more severely affected and are given the diagnosis of intellectual disability. It's important for a child to be reassessed as they age and be offered targeted help when needed.

Behaviour

Not all children or adults with a 16p11.2 microduplication 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 16p11.2 microduplication are known to have or have had delayed speech and language difficulties. Difficulties vary between children but an assessment by a speech and language therapist as well as therapy when needed is recommended.

Neurodevelopental disorders and mental health

Some children with a 16p11.2 microduplication are diagnosed with autism spectrum disorder (ASD); others have behaviours similar to those of autism but do not receive an ASD diagnosis. Some children have difficulties with attention and have been given an ADHD diagnosis. Others have anxiety or emotional difficulties. Some adults with a 16p11.2 microduplication have anxiety or depression, others have obsessive compulsive disorder (OCD) or bipolar disorder and a few people with schizophrenia have been identified. However, it is thought that neurodevelopment and mental health difficulties may develop due to additional unknown factors.