



### Why did this happen?

There is no known cause for 7q21.11 deletions. Nothing you did or did not do has caused this.

### Can it happen again?

Provided that neither parent carries the same genetic change as their child, the chance of having another affected child is extremely low – less than one percent. This residual risk is due to a situation called ‘gonadal mosaicism’ where a genetic change is present in the egg or sperm but not in any other cells of the body. None of the children reported with 7q21.11 microdeletions inherited it from a parent. Specific advice should be sought from your clinical genetics team such as a genetic counsellor.

### How many people have a 7q21.11 micro-deletion?

A 7q21.11 microdeletion is very rare. Very few patients (fewer than 10) are reported in the scientific literature. However, with increasing use of ‘next generation’ genetic diagnostic technology, we will identify more people affected by this condition. This will help us provide more useful information to families.

### Can this be cured?

There is no cure as the effects of the deletion took place during development in the womb. However, knowing the diagnosis means that appropriate monitoring and treatment can be put in place.

## Families say ...

“ Our daughter is a sweet little girl who has an overwhelmingly caring heart. She has contributed so much to our life. She blesses us daily along with some challenges along the way. But we wouldn’t want it any other way, she is a true gift. She brings so much laughter and happiness to our family. We truly are the lucky ones.”

## Inform Network Support

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This 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 guide was written by Dr. Alisdair McNeill (Honorary Consultant Clinical Geneticist) and Hollie Cowley (BSc), University of Sheffield, UK.  
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# 7q21.11 microdeletions



## What is a 7q21.11 microdeletion?

Our bodies are made up of millions of cells, which arise from one single cell made at the time of conception. Each cell contains 46 chromosomes, arranged in 23 pairs, on which genes are arranged like beads on a string. Genes act as instructions telling our bodies how to grow, develop and function. Since chromosomes come in pairs, genes also come in pairs. Each parent passes on only one copy of each pair of chromosomes so that the baby has a new set of pairs of chromosomes inherited equally from father and mother.



A 7q21.11 microdeletion is a genetic condition where a small segment of chromosome 7 is lost. Each chromosome has a long (q) arm and a short (p) arm. A 7q21.11 microdeletion occurs on the long arm of chromosome 7 at band 21.11 (see diagram).

A deletion in a chromosome can disrupt many surrounding genes. Although there are only a few reported cases of a microdeletion at 7q21.11, it is thought that the main associated genes are *CACNA2D1* and *PCLO*. They play an important role in sending signals in the brain, telling our bodies what to do.

## Families say ...

“ A major developmental milestone was when she turned 2 years 6 months and began to walk. Not too far after it seemed like her speech and learning improved to the next level. At age six she is also in swimming lessons where she can hold her breath under water and begin to front crawl with assistance. Although she struggles with learning in the classroom, she continues to meet her individualized goals and makes progress each year. ”

## Consistent Features:

- Global developmental delay
- Intellectual disability, usually including a speech delay

### Other possible features include:

- Epileptic seizures
- Difficulties with fine motor skills
- Behavioural concerns, including autistic traits and challenging behaviours.

*These features are not specific for 7q21.11 microdeletions. Diagnosis must be made with a specific genetic test.*

## Medical concerns

### ■ Seizures/Epilepsy

Some children experience generalised seizures, although they are not a consistent feature. Generalised seizures affect the whole brain and happen when there is a sudden burst of electrical activity in the brain.

### ■ Alterations to brain structure

Three children had alterations to brain structure, which were identified by brain scans. A brain scan in a fourth child was normal. These alterations in brain structure may contribute to the development of epilepsy, and might make treating the epilepsy more difficult.

## Management:

- EEG (measurement of the brain's electrical activity) if seizures are suspected.
- Brain imaging with MRI (if a child has seizures or limb weakness)
- Follow up by a developmental paediatrician or community paediatrician
- Physiotherapy and speech and language therapy as needed
- Genetic counselling as needed to provide support and advice about the genetic condition

## Development

### ■ Physical development

All children with a 7q21.11 microdeletion showed signs of developmental delay and some showed general motor difficulties. Most children were able to walk at around 18-24 months; however, one child was not able to walk until 3 years of age. One child struggled to walk long distances at the age of 15.

### ■ Learning

All reported children with the condition had significant difficulties with learning and showed a below average IQ for their age. Most attended a special education needs school.

### ■ Behaviour

Some children with this condition display autistic traits or challenging behaviour. One child showed symptoms of hyperactivity at 4 years of age. A six-year-old girl demonstrated stereotyped movements, sensory issues and mild anxiety.

### ■ Speech

Children typically experience speech and language delay to a varying degree. One child at the age of six had no recognisable words and another could not speak in sentences at the age of 15.

### ■ Fine motor skills

Some children may experience difficulties with holding and using everyday items such as cutlery.

### ■ Growth

Children with 7q21.11 microdeletions are generally of normal birth weight, with height and head circumference measurement that show no significant changes from the average.

“ Our daughter has difficulty being comfortable with clothes, which involves changing clothes multiple times a day and creates behaviour issues when trying to get ready for school in the morning. At home, she likes to get under a blanket to relax and unwind. ”