How common is ring chromosome 17?

Ring chromosome 17 is very rare. So far (2021) less than 30 people with this genetic diagnosis have been reported in the medical literature. Others will have a ring chromosome 17 but are not reported in the medical literature. It is expected that more people will be diagnosed with this condition as genetic testing becomes more routine but it is expected to remain rare.

Why did this happen?

When children are conceived, the genetic material is copied in the egg and sperm that make a new child. During this stage of 'biological copying' random rare changes occur in the genetic code of children that are not seen in the DNA of their parents. This happens naturally and is not due to any lifestyle, dietary or environmental factors. No one is to blame and nobody is at fault. Such changes happen to everyone but it's only when a change affects an important gene (or a number of genes) that development and/or health are affected.

In most children reported with a ring chromosome 17 so far, the ring chromosome occurred by chance in that child (this is known as *de novo*) and was not found in either parent. A few children are known to have inherited a ring chromosome 17 from an un affected or mildly affected parent with ring chromosome mosaicism.

Can it happen again?

The chance of having another child affected by a rare chromosome disorder depends on the genetic code of the parents. If the ring chromosome has been shown to be *de novo*, that means neither parent was found to carry it, the chance of having another child with this chromosome is low, but still possible if a parent is found to be mosaic for ring chromosome 17 or has a specific and balanced translocation involving this chromosome (a balanced translocation is when a piece of one chromosome swaps places with that of another, but no genetic material is lost or gained). A clinical geneticist can give you specific advice for your family.

Can it be cured?

Ring chromosome 17 cannot be cured, however, knowing the diagnosis means that appropriate monitoring and treatment for many symptoms, such as seizures, can be put in place.

Families say ...

"He is 25 years old, and has supernumerary ring x and ring 17 chromosomes. He has most, but not all of the attributes listed under "ring chromosome 17 that does not involve the MCDR." He loves to read, watch TV and go on the internet. He loves to travel, has a great sense of humour and an incredible memory for facts. He constantly surprises us with what he can do."

"He is 30 years old and his straight forward interactions endear him to those he meets. He loves playing video games, building Lego, reading books and telling himself stories. He delights in playing tricks on his dad and adores animals. He's taught us a lot about doing life on your own terms, speaking truthfully and love!"

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

Websites, Facebook groups and other links:

Miller-Dieker syndrome support group:
https://www.facebook.com/groups/147617675354675

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!

Rare Chromosome Disorder Support Group
Registered in England and Wales

Charity Number 1110661
Company Number 5460413

Understanding Chromosome & Gene Disorders

Ring chromosome 17

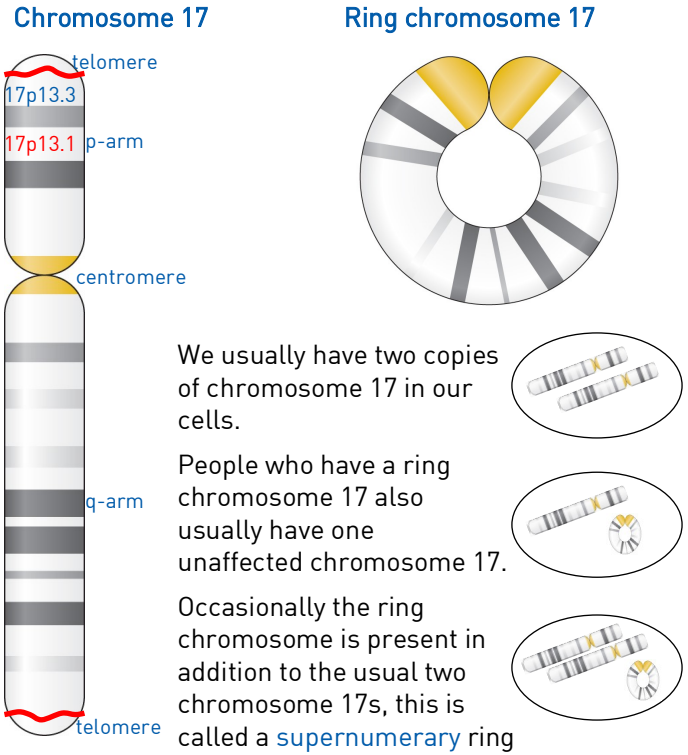
rarechromo.org

What is ring chromosome 17 syndrome?

Ring chromosome 17 syndrome is a rare genetic condition that can cause developmental delay and affect a child's learning abilities. It has also been strongly associated with epilepsy. As is common with genetic conditions, each person is affected differently and this can depend on what and how much genetic material is lost when the ring chromosome forms and if the ring chromosome is found in all of their cells.

What causes ring chromosome 17?

Ring chromosome 17 occurs when a small amount of genetic material is lost from the ends of chromosome 17, and the chromosome bends to form a closed ring instead of it's usual bar like structure.



Some people have mosaic ring chromosome 17 which means that not all of their cells contain the ring chromosome.

Ring chromosome 17 features

How people are affected by any genetic condition varies for many reasons and is greatly influenced by each person's own unique genetic makeup. For ring chromosome 17, symptoms and features also depend on what and how much genetic material is lost (if any), the activity of genes near the point of fusion, or in different tissues,and if the ring chromosome is mosaic or supernumerary.

However, the most important consideration is whether a specific piece of genetic material from the end of the p-arm of chromosome 17 is deleted. This piece of genetic material is called the Miller-Dieker critical region (MDCR) and is found in a region called 17p13.3. When the MDCR region is lost, the exterior part of the brain does not develop it's usual grooves and folds and instead appears to be smooth; this is known as lissencephaly. Children with this brain structure have severe physical and mental developmental delays, intellectual disability, and seizures. They will be given the diagnosis of Miller-Dieker syndrome.

If the ring chromosome forms and the MDCR is not lost (or more precisely, the PAFAH1B1 and/or YWHAE genes are not deleted), features and symptoms are milder and depend on which other genes are affected.

Medical and developmental concerns

It is not possible to provide a clear description of expected symptoms and features for people with ring chromosome 17 since each person can have a different amount of (or no) chromosome missing from (or duplicated at) the end of the p and/or q arm as well as a varying degree of (or no) mosaicism. In addition, so few people have been described in the medical literature. The following is just a brief summary of the main symptoms and features that have been reported so far. It's important to recognise that each child is very different and is likely to have different abilities and difficulties depending on their own Unique genome.

Four people have also been reported to have a ring chromosome 17 that involves the deletion of a gene called TP53 in region 17p13.1. These people also have chronic lymphocytic leukemia (CLL).

Children with ring chromosome 17 that does not involve the MDCR

Only thirteen people have been reported in the medical literature (2021) as having a ring chromosome 17 that does not involve the MDCR region. Varying levels of learning difficulties are mentioned as well as intellectual disability, but one child was described as having normal intelligence (as is one Unique member). Features can include:

- Developmental delay
- Learning difficulties/disorders with possible intellectual disability (ID) or cognitive function within the normal range
- Café-au-lait spots (indicating mosaicism)
- Speech and language delay or loss
- Weak muscle tone (hypotonia)
- Seizures and epilepsy
- Eye anomaly: retinal flecks
- Short stature

Children with a ring chromosome 17 that involves the MDCR can have:

- Brain anomaly (lissencephaly)
- Developmental delay (severe)
- Intellectual disability (ID)
- Seizures and epilepsy
- Small head (microcephaly)
- A small jaw (micrognathia)
- Feeding and swallowing difficulties
- Speech and language delay or loss
- Weak muscle tone (hypotonia)
- Muscle stiffness (spasticity)
- Short stature
- Excessive amounts of amniotic fluid during pregnancy (polyhydramnios)

Sadly, ring chromosome 17s that include the MDCR are also associated with a reduced life expectancy.

Management recommendations:

Children with a ring chromosome 17 should be under the care of a multidisciplinary team including a geneticist, paediatrician, neurologist and an epilepsy specialist. Children may benefit from speech and language therapy as well as physio– and occupational therapy and periodic evaluations by a developmental specialist.