Why did this happen?

Many ring chromosomes are sporadic, that is, they arise out of the blue (*de novo*). The actual cause is not known and should be regarded as an accident that happened during cell division while the egg or sperm cells were being created or after conception (when a baby is made). These accidents are not uncommon and affect children from all parts of the world and from all types of background. They also happen naturally in plants and animals. So there is no reason to suggest that your lifestyle or anything that you did caused the ring to form.

A ring 21 chromosome may also be inherited from a parent, in most cases the mother, as ring chromosomes appear to be associated with reduced fertility in men. A ring chromosome passed down in this way may stay unchanged (stable) or it can change.

Will a child with ring 21 passed on by an unaffected parent also be unaffected?

This is very difficult to answer. In general, a stable ring is less likely to cause symptoms. Signs that the ring is stable include:

- Ring stability in the parent, who is unaffected
- Ring stability in other members of the same family who are also unaffected

Sometimes, though, an apparently stable ring can cause symptoms in a baby, so it is not possible to know with certainty from chromosome tests alone whether a baby is affected or not.

Can it happen again?

So long as tests show that parents' chromosomes are normal, they are very unlikely to have another affected child. All the same, you should have a chance to discuss prenatal diagnosis if you would like it for reassurance.

How common is ring 21?

Generally speaking, ring chromosomes occur in around one in every 25,000 known conceptions. At the time of writing, *Unique* had a small community of 23 member families with a ring 21. Families who wish to do so can make contact with each other.

Inform Network Support

Understanding chromosome disorders Unique

Rare Chromosome Disorder Support Group,
G1 The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK
Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

When you are ready for more information, *Unique* may be able to help. We can answer individual queries and we also publish a detailed leaflet about ring 21.

This leaflet 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. The information is believed to be the best available at the time of publication and the content of the full leaflet on which this text was based was verified by Dr Nicky Foulds, consultant clinical geneticist, Wessex Clinical Genetics Service, 2004 and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

Rare Chromosome Disorder Support Group
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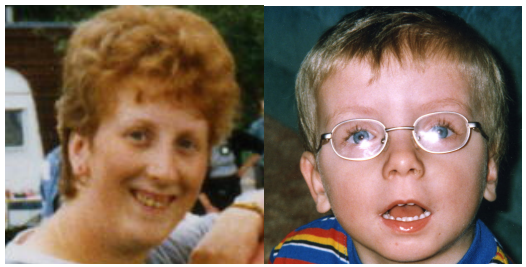
Charity Number 1110661
Company Number 5460413

Understanding chromosome disorders Unique

Ring 21

rarechromo.org

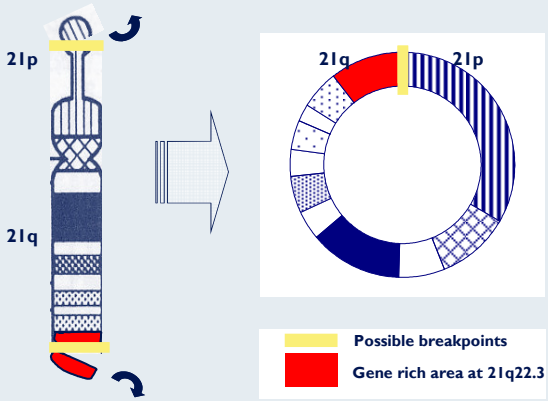


What is ring 21?

Ring 21 is a rare genetic condition caused by having one chromosome 21 that has formed a ring. The other chromosome 21 is generally intact and has the normal form of a chromosome.

Chromosomes, which contain our genetic information, are usually rod-like structures. In human beings there are 46 chromosomes, occurring as 23 pairs. Chromosomes 1 to 22 are numbered approximately from longest to shortest, although chromosome 21 is in fact the smallest chromosome and holds no more than 200 to 400 of the total of around 30,000 genes. The 23rd pair are the sex chromosomes, two Xs for females and an X and a Y for males.

Each chromosome has a short arm ('p' for petit) and a long arm ('q'), separated by a pinched area known as the centromere. Chromosome 21 has a very small short arm that does not contain genes relevant to development. The ends of the chromosome are called the telomeres.



How does the ring form?

For reasons that are not understood, the ends of the chromosome may simply join up. This is known as telomere-to-telomere fusion. More often, the ends of the chromosome break off and the broken, 'sticky' ends fuse at the breakage points. The broken fragments are lost, and with them any genes they may contain. With a ring chromosome 21, only the genes on the long arm matter. The ring can also be caused by part of the long arm forming a loop, joining up and breaking off.

Different types of ring 21

Some people with ring 21 chromosomes are healthy and develop normally. Their unusual chromosomes are discovered by chance, during tests for infertility or repeated miscarriages or after having an affected baby. In other people the ring 21 chromosome affects development and learning and can also cause medical problems.

Type I

Discovered by chance, *or* during investigations for infertility or repeated miscarriages *or* after a baby with abnormalities or Down's syndrome is born

- Normal development
- No effects on health
- Possibly short stature
- In boys and men, puberty may possibly be slightly delayed
- The ring makes no difference until people want to have children

It is believed that people who show no effects of their ring chromosome have a single ring with a break at the very end of the long arm of chromosome 21 in the band known as 21q22.3.

Type II

The effects of this type of ring 21 range from slight to severe and can vary between different members of the same family. The reason for these differences is not fully understood. Some of the more common effects are:

- Short stature
- Small head circumference (microcephaly)
- A variable degree of learning difficulty or disability
- Vulnerability to infection
- Possibly, increased vulnerability to seizures

Type III

A few people with ring 21 have:

- Features similar to Down's syndrome

Down's syndrome is caused by having three copies of a chromosome segment known as the 'Down's critical region' at 21q22.3. People with ring 21 and Down's syndrome are believed to have a double-sized or multi-sized ring in many cells, so that the 'Down's critical region' appears at least three times in most cells.

Development

Growth

Ring chromosomes usually give rise to short stature. This is common with ring 21 but does not affect everyone. Most babies born small grow into short children but this, too, is not always the case.

Learning

People unaffected by their ring chromosome would not normally have any learning difficulty or disability caused by the chromosome disorder. Other people with ring 21 would expect to need a degree of learning support, but this may be slight. Youngsters with the Down's presentation of ring 21 are likely to need more support.

Sitting, moving …

Babies may be delayed in reaching their milestones of rolling, sitting and walking but go on to become mobile and independent. Others become mobile with support, but for a few this level of mobility is not possible.

Behaviour

We are not aware of a specific type of behaviour linked with ring 21, but our experience is that a high level of activity is a common feature.

Medical concerns

High rate of infections

As a group, children are vulnerable to infection and may also show delayed wound healing. Respiratory and sinus infections are especially common and bacterial infections are common in childhood. Some children have low levels of antibodies (IgA, IgG). Your child's doctors can suggest sensible steps to protect your child against infection and may recommend enhanced antibiotic cover.

Heart

Your baby will have a thorough heart exam and further cardiac evaluations if any abnormal heart sounds are heard. A variety of structural heart abnormalities have been found in children with ring 21. Some will resolve naturally on their own while others are corrected by surgery.

Blood disorders

Some children with a deletion involving 21q22 may have wounds that are slow to heal and bruise easily.