

Ring chromosomes

The most common features among people with a ring chromosome are:

- **Slow growth and eventual short height**
- **Some level of learning difficulty or disability**
- **Very small head (microcephaly)**
- **Some unusual facial features. These can be subtle or more obvious**
- **Anomalies in skin colouring, such as streaky pigmentation**

Why did this happen?

The great majority - 99% - of ring chromosomes are sporadic, that is, they occur out of the blue. The actual cause is not known and should be regarded as an accident that happened in cell division in the process of making sperm or egg cells. 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. There is no reason to suggest that your lifestyle or anything that you did caused the ring to form.

Very occasionally, a ring chromosome may be inherited from a parent. This can occur when the parent has the ring 18 in all their cells or when they have neighbouring cell populations, some with the ring chromosome and others with normal chromosomes. This is known as mosaicism. In most (90%) of familial cases the ring has been inherited from the mother, as ring chromosomes appear to be associated with reduced fertility in men.

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.



Ring chromosome 18

Inform Network Support



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Unique mentions other organisations' message boards and websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it.

When you are ready for more information, Unique can help. We can answer individual queries and we also publish a more detailed leaflet about ring 18.

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 is derived from the full text leaflet, verified by Dr Jannine Cody, president of the Chromosome 18 Registry November 2004 and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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Registered in England and Wales

Charity Number 1110661
Company Number 5460413

Understanding
chromosome
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Ring 18

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What is ring 18?

Ring chromosome 18 is a rare genetic condition caused by having one chromosome 18 that has formed a ring.

Genetic information is contained in each cell of the body in the genetic material DNA, tightly coiled and forming rod-like structures called chromosomes. In human beings there are 46 chromosomes, occurring as 23 pairs. Chromosomes 1 to 22 are numbered approximately from longest to shortest and the last 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.

In people with ring chromosome 18, one chromosome 18 is usually intact but the other has formed a ring. When a ring forms, both arms of a chromosome break and the broken 'sticky' ends fuse at the breakage points. The broken fragments are lost, and with them any genes they may contain.

Development works with clockwork precision and the right amount of genetic material is needed for normal growth and development. If any genetic material is missing or extra, then it is likely to result in some problems.

Specific features of ring 18

These vary between individuals and depend partly on how much material has been lost from each arm.

More common in people who have lost the end of 18q (18q-)

- Very narrow or closed ear canals
- Structural heart abnormality
- High arched or cleft palate
- Unusual feet or hands
- In boys, minor genital abnormalities, such as the hole normally at the end of the penis being beneath the shaft (hypospadias)

Development

■ Growth

Children are usually short and become short adults. Without treatment, predicted adult height is around five foot (150cm).

■ Learning

Most people with ring 18 will need support for their learning difficulties, but the range is very broad. The majority seem to have a moderate learning disability and a few have a severe disability. A few people will learn at a speed within the normal range.

■ Speech

Speech is specifically delayed but eventually most children acquire speech and some talk fluently. Contributory factors include hearing loss (see below), difficulties with articulation due to a cleft or very high palate (roof of the mouth) and any learning difficulties. Children generally benefit from speech therapy to help develop and coordinate the action of the muscles used in speech.

■ Sitting, moving ...

Generally, babies are late to roll over and sit and children are late to walk. Our experience is that babies learned to sit at 7-18 months and took their first steps at 18 months - 5 years. Many children have low muscle tone. This generally improves with age and practice and children benefit from regular physiotherapy and occupational therapy.

More common in people who have lost the end of 18p (18p-)

- Ptosis (drooping of upper eyelids)
- Holoprosencephaly. This is a developmental defect that occurs when the brain and face are forming. The brain does not grow forward and divide into two hemispheres as it is supposed to do. The severity varies, with the least affected people having some fused structures and the most severely affected having a single brain mass instead of two separate lobes. Most people come between these two extremes. The middle of the face can also be affected: a mildest effect can be a single front tooth; a severe effect, a single eye.

Medical concerns

■ Infections

Some children are prone to catching infections and these can be quite severe. Some children have low antibody (IgA) levels. IgA plays a key role in defending the body against infection that invades through the mucous membranes, such as the nose, eyes, lungs and intestines. It is found in breast milk. Your child's doctors will advise on sensible steps to take to protect against infection.

■ Heart

As losing the end of 18q can be associated with heart conditions, all children should have a thorough heart exam and detailed investigations if abnormal heart sounds are heard. Some heart abnormalities may resolve naturally in time and others can be corrected with surgery. Children generally thrive after heart surgery.

■ Hearing

Some children with loss from 18q have very narrow or blocked ear canals. The severity varies between individuals and in some cases the canals can be widened and straightened. In many children, there is an associated hearing loss which can be usually be helped by wearing a surgically implanted (bone anchored) hearing aid.

■ Eyelids and vision

Some babies cannot fully open their upper eyelids (ptosis). Both eyes are usually affected and this is evident from birth. If ptosis stops your child from seeing properly, it can be corrected in a one or two-stage surgical operation.

■ Palate

The roof of the mouth may not have joined fully, leaving a gap (cleft) in part of the palate. In some babies the palate is intact but very high-arched. Both conditions affect feeding and speaking. You should be offered feeding support and your baby may feed better with special feeders. A cleft palate is usually repaired surgically before your baby's first birthday.

■ Genitals

Occasionally, baby boys are born with minor abnormalities of the genital area such as hypospadias. If necessary, surgery can achieve a more normal urinating position.

■ Feet and hands

A baby's hands and feet may occasionally be unusually formed. This should not generally affect hand function. Surgery can be helpful to achieve the best possible position and function of your child's feet for walking.