What are the consequences of a ring chromosome?

Regardless of which chromosome is involved, a ring chromosome has similar effects. The most common effects 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
- Some patchy or streaky skin colouring, usually café au lait coloured

Why did this happen?

The great majority - 99% - of ring chromosomes are sporadic, that is, they happen out of the blue. The cause is not known and they 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 can 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.

Very occasionally, a ring chromosome may be inherited from a parent but this has never been reported with ring 2.

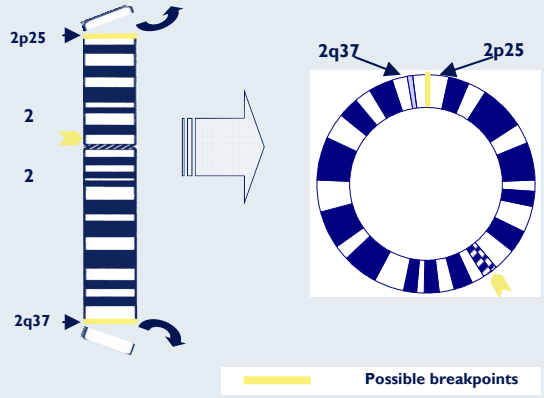
Can it happen again?

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 2?

In 2005, only 11 children with this unusual chromosome disorder had been described in the medical literature and *Unique* had five affected member families. Families within *Unique* can make contact with each other if they feel this would be helpful.

Possible formation of a Ring 2 chromosome



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* can help. We can answer individual queries and we also publish a more detailed leaflet about the effects of ring 2.

This information sheet 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 Fowzan Alkuraya, Fellow, Genetics & Metabolism, Children's Hospital, Boston & Harvard Medical School, US and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK, 2005.

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Ring 2

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What is ring 2?

Ring 2 syndrome is an extremely rare chromosome disorder caused by the presence of a ring chromosome in the cells of the body.

Chromosomes, the structures inside the nucleus of each cell that contain our genetic information, are usually thread-shaped. For reasons that are not understood, the ends of a chromosome sometimes join up to form a ring. Any one of the 22 different non-sex chromosomes can form a ring and so can either of the sex chromosomes X and Y. This seems to occur very rarely indeed to chromosome 2, which is almost the largest chromosome.

When a ring chromosome forms, either the tips fuse with no loss of genes or chromosome material, or the chromosome breaks at each end and the 'sticky' broken ends join to form a ring. The broken fragments are lost, and with them any genes they may contain.

Features in ring 2

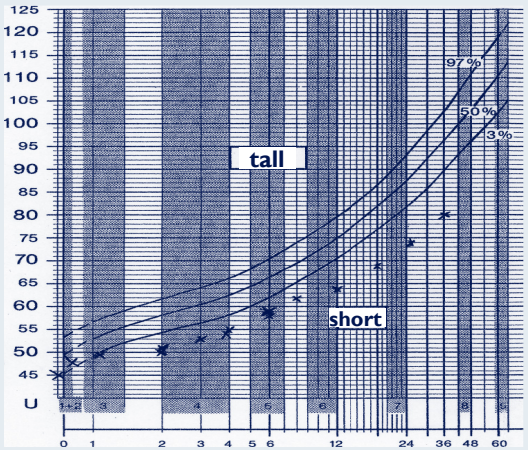
In addition to the general consequences of a ring chromosome (see overleaf), there may be specific effects of ring 2. In almost all children known so far where genes have been lost, the short arm of chromosome 2 has broken at band p25 and the long arm has broken at band q37. A deletion (loss) from chromosome 2q37 is more common than ring 2. Most of the features in the list below have also been found in some children with 2q37 deletions.

- Feeding difficulties in babies and young children, usually minor.
- Hypotonia (showing as floppiness) in babyhood and sometimes in early childhood.
- Hernias, especially umbilical and inguinal (in the groin).
- Vulnerability to behaviour difficulties, including autism.
- Heart conditions.
- Small genitals, occasionally with minor anomalies.
- Usually minor foot anomalies.
- Loose, flexible joints.

Development

Growth

Growth delay is usually obvious during pregnancy and continues after birth, with the typical growth rate below the lowest curves on a growth chart. Children may be evaluated for growth hormone deficiency and treated if appropriate.



Height in a child with ring 2 from birth to 3 years. Measurements in centimetres.

Sitting, moving, walking

Any delay in reaching the typical baby developmental milestones seems to be usually slight. The *Unique* experience is that babies rolled over at 4-10 months, sat alone at 8-18 months, became mobile at 13-15 months and walked at12-25 months.

Learning

Children can face a broad spread of ability to learn. Some children are apparently unaffected cognitively but others may have severe and complex learning needs.

Speech and communication

There is little evidence. However, in one child studied to the age of 10, speech improved noticeably with therapy. Speech therapy should therefore be available.

Behaviour

From the little information available, it appears that some children may have a high level of activity and may develop challenging behaviour. Families should have ready access to behaviour management support.

Medical concerns

Many babies and children with ring 2 are perfectly healthy. Some medical conditions and abnormalities appear to be more common in children with ring 2, so the paediatricians who care for your baby's health will be especially alert for these.

Heart conditions

Babies will have a careful heart exam and a detailed cardiac review if any abnormal heart sounds are heard. Structural abnormalities of the heart are known to occur fairly commonly in children with a small deletion from the end of the long arm of chromosome 2 (2q37 deletions) and may be even more common in babies with ring 2 syndrome. Examples include holes between the lower chambers of the heart (ventricular septal defects), persistent ductus arteriosus (persisting structure of the fetal heart), coarctation (narrowing) or displacement of the aorta, the blood vessel that leads from the heart to the body. While some conditions resolve naturally with time, a few babies will need surgery. Children generally thrive after surgery.

Feet and limbs

Feet may be unusually positioned or formed. Examples include rocker bottom feet (the sole is curved like a rocker), club foot (talipes), flat feet and a tendency for the feet to roll outwards or inwards. Stretching and physiotherapy will usually bring some improvement but surgery may be needed.

Genitals

Babies may have minor anomalies in the genital area. In boys, the genitals may be small. If the testicles have not descended into the scrotum at birth, they may descend later; if not, they can be brought down in a surgical operation called orchidopexy.

Bones and skeleton

Genes near the end of the long arm of chromosome 2 are known to be important for the development of the bones and skeleton and people with a 2q37 deletion are susceptible to a condition known as Albright's hereditary osteodystrophy (AHO)-like syndrome. AHO is a genetic condition that affects the way the body lays down calcium. It is not known yet whether individuals with ring 2 are also susceptible but it is considered likely.