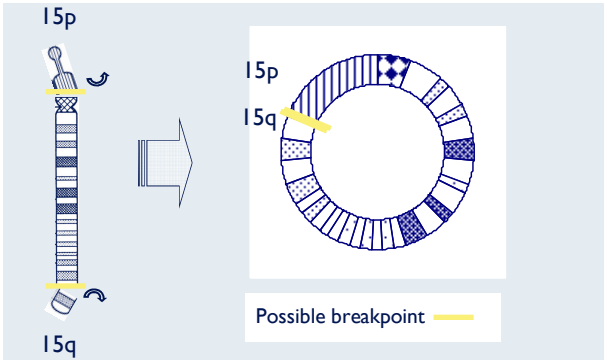
Formation of a ring 15 chromosome



Why did this happen?

The great majority - 99% - of ring chromosomes are sporadic. The 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 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 15 may be inherited from a parent. In most 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.

There is a less common type of ring chromosome 15 that occurs when a third chromosome 15 is present as a ring. This is called supernumerary (extra) ring 15 and the features are different to those of the ring 15 disorder described in this leaflet.

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

Rare Chromosome Disorder Support Group
Registered in England and Wales
Charity Number 1110661
Company Number 5460413

Understanding chromosome disorders Unique

Ring 15

rarechromo.org

What is ring chromosome 15?

Ring chromosome 15 is a rare genetic condition caused by having one abnormal chromosome 15 that forms a ring. Genetic information is contained in each cell of the body in the genetic material DNA, tightly coiled and formed into 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.

Chromosome 15, along with chromosomes 13, 14, 21 and 22, has a very small short arm that does not contain genes that are relevant to development. This group of chromosomes is known as 'acrocentric' chromosomes, meaning that the centromere is near one end.

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, it is likely to result in problems in growth and development and most often also in the functioning of the body systems.

Features of people with a ring chromosome

The most common features are:

- Slow growth and eventual short height
- Some level of learning difficulty or disability
- Very small head (microcephaly)
- Unusual facial features including a triangular face. These can be subtle or more obvious
- Patchy or streaky skin colouring

Other features

Other features vary between individuals. They depend partly on how much material has been lost from the long arm of chromosome 15.

- In boys, undescended testes at birth or hypospadias (the hole is under the penis instead of at the end)
- Heart conditions
- Club foot (talipes)
- Floppiness (low muscle tone, hypotonia)

Development

Learning

Most children are believed to need a moderate level of learning support but some are only slightly affected, others may not be affected at all and others may be severely affected. Generally memory is reported as good, but abstract thinking and mathematical skills are underdeveloped. Many children will learn to read and write but this will not be possible for all.

Speaking

Children usually speak late and may have specific difficulties in tongue movement as well as learning difficulties. In this case, they may learn to communicate well by signing before their speech develops.

Sitting, moving …

Most babies and children are somewhat delayed in learning to move, sitting on average around 11 months and walking around 22 months. Babies may be unusually floppy (hypotonia, also called low muscle tone). Physiotherapy and occupational therapy are helpful.

Behaviour

Most people with ring 15 appear to be pleasant, co-operative and sociable and mix well in social situations. Parents will still benefit from guidance to maintain good behaviour, teach clear limits and support them if behaviour becomes negative.

Growth

Babies, children and adults with ring chromosome 15 are small. Everything about them is small, including their head, but their hands and feet may be especially tiny. There is a gene near the end of the long arm of chromosome 15 that affects growth and losing it probably makes children shorter.

Medical concerns

Most people with ring 15 do not have serious medical conditions and there are many reports in the medical literature of adults. These are the more common concerns that have been reported.

Heart

Babies will have a thorough heart exam and further tests if a heart murmur or abnormal heart sounds are heard.

A minority of babies are born with a structural heart abnormality. These are most commonly small holes in the muscular wall between the chambers of the heart. Small holes may resolve naturally with time but others need to be corrected with surgery. Children usually thrive after surgery.

Kidneys

Babies may also have an ultrasound scan of their kidneys to detect any defects in the kidneys themselves or the drainage system through the bladder.

Genitals

At birth, the testes may not have come down into the scrotum in boys. If they remain within the abdomen, the testes can be brought down and anchored. Some boys may be born with the hole usually at the end of the penis on the underside instead. Surgery may help to achieve a more normal urinating position.

Feet and hands

Your baby's hands and feet will be carefully examined at birth. Some anomalies do not need treatment – such as incurving fifth fingers – but babies with club foot (talipes) will be carefully assessed to decide the best treatment to enable a good walking position.

Joints

Babies' joints will be carefully assessed, in particular the hip joints as they may develop with an unusually shallow socket. The socket can normally be improved with splinting and if necessary surgery and immobilisation in a plaster cast.

Hearing and eyesight

There is little formal information but within *Unique*, most children have normal hearing and those with a hearing impairment have glue ear which causes a temporary loss as a result of frequent ear infections. Eyesight does not appear to be generally affected.