## Why did this happen?

Ninety-nine per cent of ring chromosomes are sporadic, that is, they occur out of the blue. 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. They affect children from all parts of the world and from all types of background. They also occur 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, most often from the mother. To date only one mother: daughter pair with a ring chromosome 13 has been described in the medical literature and both are very mildly affected.

## Can it happen again?

So long as tests show that the 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.

**Inform Network Support**

Understanding chromosome disorders Unique

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When you are ready for more information, *Unique* may be able to help. We can answer individual queries and we also publish a more detailed leaflet about ring 13.

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 Sixto Garcia-Minaur, clinical geneticist, North West Thames Regional Genetics Service and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.



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# Ring 13

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## What is ring 13?

Ring chromosome 13 is a rare genetic condition caused by having an abnormal chromosome 13 that forms a ring.

Chromosomes are the structures in the nucleus of the body's cells that carry genetic information. There are 23 pairs of chromosomes, one member of each pair having come from the mother and the other from the father. The chromosomes are numbered from 1 to 22 according to size and the 23rd pair are the sex chromosomes, usually two Xs for a girl and an X and a Y for a boy. Chromosomes are usually rod-shaped and each one has a short (p) arm and a long (q) arm.

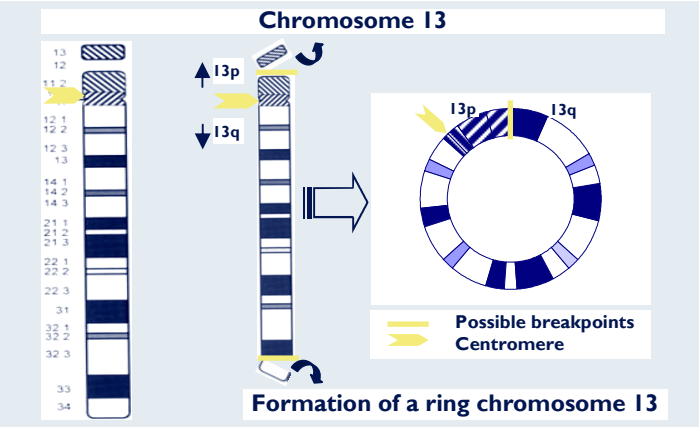
In people with ring chromosome 13, one chromosome 13 is the usual shape but the other has formed a ring. When a ring forms, both arms of a chromosome break and the broken 'sticky' ends fuse at the breakage points. The broken fragments are lost, and with them any genes they may contain. With a ring chromosome 13, only the genes in the long arm matter.

### What are the consequences of having a ring chromosome?

A ring chromosome tends to have similar effects, regardless of which chromosome is involved. 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
- Patches or streaks of darker or lighter skin

The effects of the ring are altered by cells with a different chromosome make-up (mosaicism). The chromosomes in blood cells may not reflect precisely the chromosome make-up of cells in other body tissues and this makes it more difficult to make accurate predictions for a particular child.



Formation of a ring chromosome 13

### Does it help to know the exact breakpoints in the ring?

It can help to predict the severity of problems. If the breakpoint is closer to the end of the long arm (eg at 13q34, *above*), a smaller amount of material and fewer genes will be lost, and this will have fewer consequences. If the breakpoint lies higher up (eg at13q22), more chromosome material will be lost and this will have more far-reaching consequences.

### Other features of ring 13

Depending on what chromosome material and genes have been lost, the most common additional features of ring 13 syndrome are:

- Unusual formation and position of one or both feet.
- High palate (roof of the mouth). More unusually, it may be cleft (split).
- Unusual hands. Fingers may be short. Thumbs may be unusually formed or in an unusual position.
- Unusual position or formation of the anus (bottom). It may be covered with skin, very small or unformed.
- Incorrect position for outlets to the urinary system. This can usually be corrected with surgery.
- Some degree of genital abnormality in boys. This may be minor or more problematic.
- Small lower jaw.
- Kidney anomalies.
- Abnormal brain structures.
- Abnormal eye structure, most obviously very small eyes.

## Development

**'Happy, friendly and helpful'**

### Growth

Typically, babies grow slowly in the womb, have a low birth weight, and have difficulty in putting on weight afterwards. Slow growth is especially obvious in babies with a small ring and a large deletion. Some children with a breakpoint at 13q32 or 13q34 follow a growth pattern that is closer to average. There is little information on eventual height but the known range is five foot (150 cm) to five foot nine inches (175 cm).

### Learning

Ring 13 will usually affect the speed at which children learn and set some limits on their eventual achievement. Just how slight or far reaching the effects are will become clearer as your child develops, but the possible range is really very broad. It is not usually possible to predict precisely from the chromosomes what the effects on learning will be, but in time it will become easier to suggest the outlook from the chromosomes and observations of your child's development.

### Speech and communication

Speech and language delay can be expected in children with ring 13 and in the most mildly affected children may be the first sign of the disorder. In general, the level of speech difficulty reflects the level of learning difficulty. Some youngsters will acquire speech which may be quite fluent, while for a few speech will not be possible.

### Mobility and activity

Some degree of delay in reaching mobility milestones for sitting and walking is very common but the degree of delay is variable. Many babies have abnormal muscle tone – typically their muscles are too floppy (hypotonia). Physiotherapy is helpful and some children may need supports to help them to walk. All adults within *Unique* were able to walk indoors and out at some point, but a few relied on a wheelchair outdoors.