

## Why?

Ninety-nine times out of 100, a ring chromosome occurs out of the blue. There was nothing you did that caused it and, although this is hard to accept, nothing you could have done to prevent it either. Very occasionally a mother who is unaware that she has Ring 14 passes the chromosome on to her child – but again without knowing, she couldn't prevent this. Assisted reproduction offers no guarantees either: Ring 14 has been reported after treatment for infertility just as it has through normal reproduction.

## Can this happen again?

The chances of having another affected child depend on the results of chromosome tests on the parents but it is usually extremely unlikely that you would have another child with Ring 14 syndrome. However, in the very rare cases where the mother herself has the same ring, there is a real possibility of having another affected pregnancy. Each family is unique so you should have a personal interview to discuss the findings of the chromosome tests with a geneticist or genetic counsellor.

## How is Ring 14 diagnosed?

Cells from a blood sample are treated so that the chromosomes can be examined under a high resolution microscope. The ring shape of one copy of chromosome 14 is usually quite obvious. Molecular tests can be carried out in research laboratories to show exactly what material is missing, but at present this rarely helps in predicting the effects.

## Inform Network Support



**Rare Chromosome Disorder Support Group,**  
G1 The Stables, Station Road West, Oxted, Surrey RH8 9EE, UK  
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**Ring 14 International** is a support group for families affected by Ring 14 syndrome. The centre is in Italy but it operates worldwide and aims not only to disseminate information but also to promote and fund research.  
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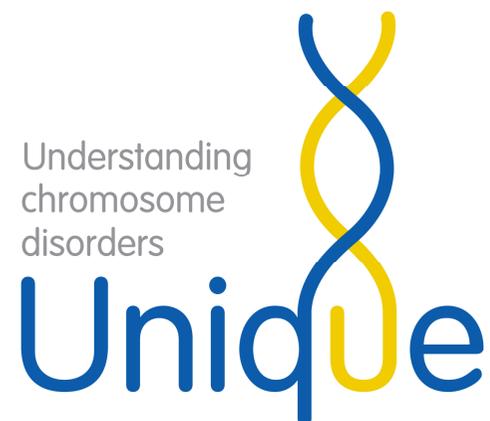
When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a full leaflet about the effects of Ring 14.

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 medical content has been verified by Dr Melanie Manning, clinical instructor, Stanford University School of Medicine, and by Unique's Chief Medical Adviser 2004

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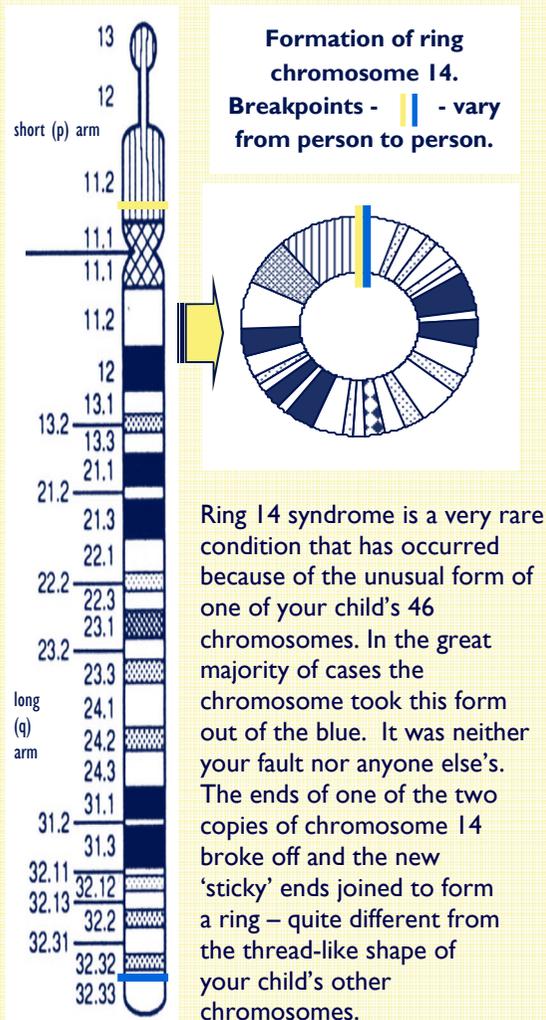


# Ring 14 syndrome

[rarechromo.org](http://rarechromo.org)



## What is Ring 14 syndrome?



### How common is Ring 14?

No-one knows for certain. There is a support group in Italy, a US-based website and a support network within *Unique*. In 2005, *Unique* had 25-30 members.

## Development

### ■ Motor development

Children are typically somewhat late to sit and walk. Once mobile, many become active and some develop considerable sporting ability. Others rely on support or a wheelchair for walking distances. One or two do not walk. Overall, the picture is very varied.

### ■ Learning

Some people have no learning difficulties at all, others face mild difficulties but more often they are moderate to severe. Children may learn to read and write single words and be able to tell their name and address.

### ■ Speech

Children have speech and language delay but a few of them master complex sentences and subtleties of communication. More commonly, children use isolated words and use gestures, pictures, signing and expression to communicate.

### ■ Behaviour

Although there is no specific pattern of behaviour problems linked with Ring 14, some children are restlessly active and find concentration difficult and a smaller number may react to their frustration at being unable to communicate by being challenging. Medicines taken for seizures can affect behaviour. Parents find management techniques and occasionally medication helpful.

### ■ Growth

Some children are short and many children have a small head, though this is rarely noticeable.

## Medical concerns

- **Seizures** affect the great majority of people with Ring 14. They start in early childhood, typically in the middle of the first year. A few children outgrow their seizures and some enjoy seizure-free months and even years. In others the seizures remain a frequent event.
- The range of children's antiepileptic medicines has recently grown rapidly so treatment can be fine-tuned to achieve the best possible control. Most children need a multi-drug approach to balance seizure control with side effects. In around half of the children the structure of the brain appears unaffected by Ring 14.
- **Respiratory infections** are common, especially in young children, and they can be severe. Croup, bronchitis and pneumonia are much more common than in other children especially in winter. Children should be fully immunised with the vaccines recommended locally.
- **Hypotonia** - a physical floppiness as though the body is profoundly relaxed - is common and contributes to the delay in children's physical development.
- **Retinal markings** Fleck-like white or yellow markings can be seen on many children's retinas when viewed through an ophthalmoscope. However, these do not affect eyesight.
- **Skin** Spots or patches of coffee-coloured or light skin may be visible on the body. These are harmless.

**Feeding** can pose a challenge for families faced with a child who is small for their age, has little appetite and may have difficulty both in sucking strongly and in coordinating sucking with swallowing. Reflux can usually be controlled with feed thickeners and antacid medicines. Babies with very severe reflux can be treated with surgery or in some cases a feeding tube can be inserted direct into the stomach.