**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.
**What is Ring 14 syndrome?**

Ring 14 syndrome is a very rare condition that has occurred because of the unusual form of one of your child’s 46 chromosomes. In the great majority of cases the chromosome took this form out of the blue. It was neither your fault nor anyone else’s. The ends of one of the two copies of chromosome 14 broke off and the new ‘sticky’ ends joined to form a ring – quite different from the thread-like shape of your child’s other chromosomes.

**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 restless, 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.

**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.