

Why did this happen?

The great majority - 99% - of ring chromosomes are sporadic. The actual 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 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. In most cases the ring has been inherited from the mother, as ring chromosomes appear to be associated with reduced fertility in men. However, there is at least one case on record of a father passing a ring 22 onto his child.

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.

Passing on a Ring 22 is very rare. Only four cases have been reported. In one family an unaffected father passed the ring to his daughter who had a slight learning disability. She passed it on to her twin daughters who, like their grandfather, were unaffected.

How common is Ring 22?

Details of over 100 people with Ring 22 have been mentioned in research reports or on the internet but the real total is very likely to be much higher. All the same, Ring 22 is so uncommon that most doctors and paediatricians have never met an affected child before. *Unique* knows many families with Ring 22 and currently has more than 60 affected members. Member families who would like contact can get in touch with each other.

Inform Network Support



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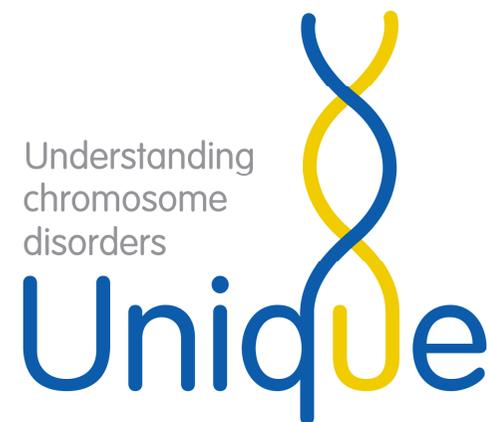
Email support group
<https://groups.yahoo.com/neo/group/ring22/info>

At Chromosome 22 Central you will find
some information on Ring 22.
www.c22c.org

Other sites are listed to be helpful to families looking for information but a listing does not imply that *Unique* endorses its content. When you are ready for more information, *Unique* can help. We can answer individual queries and we have a longer text about the effects of Ring 22. 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 Aaron Jeffries, Institute of Psychiatry, London 2005 and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK 2004.

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Ring 22

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What is Ring 22?

Ring chromosome 22 is a rare genetic condition caused by having a chromosome 22 that is formed into a ring instead of the normal rod-like structure. People with Ring 22 are usually healthy and look like everyone else, but their development and speech are delayed.

What is a chromosome?

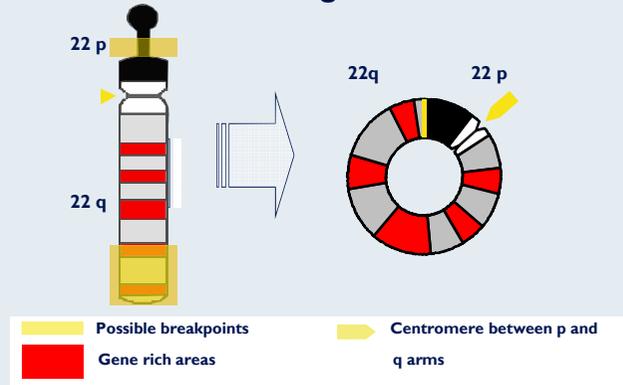
All our genetic information is contained in the cells of our body in DNA, coiled and formed into 46 rod-like structures called chromosomes. Between them, the chromosomes contain about 20-25,000 genes. Chromosomes occur as 23 pairs. We get one member of each pair from our mother in the egg, and the other member from our father in the sperm. The first 22 pairs are numbered 1 to 22, roughly according to size, so chromosome 22 is almost the smallest. The last pair are the sex chromosomes, usually two Xs for females (XX) and usually an X and a Y for males (XY). Each chromosome has a short (p) arm and a long (q) arm.

What happens when a ring chromosome forms?

When a ring forms, both arms of the chromosome break and the 'sticky' broken ends fuse. The broken fragments are lost, and with them any genes they may contain. Chromosome 22 has a very small short arm that does not contain genes that are relevant to development so it is only the long arm that matters.

A gene known as *SHANK3* is situated close to the end of the long arm of chromosome 22, and if this gene has been lost, as it usually is, most of the effects of Ring 22 will be very like the effects of 22q13 deletions seen in people with Phelan-McDermid syndrome. All the same, a child's other genes and personality, their home environment and the opportunities and experiences offered them have a vital impact on their future development, needs and achievements.

Formation of a ring chromosome 22



Development

Sitting, moving, walking

Babies reach their mobility milestones late, usually sitting alone by 24 months, crawling between 7 and 36 months and walking in their third year (range 13 months to 8 years). Many have strikingly floppy muscles (hypotonia) and this contributes to the delay. Physiotherapy is helpful and once on the move, many children overcome their slow start and any later problems with balance and some become good walkers, swimmers and cyclists.

Speech

Speech develops late. First words appear to emerge on average around the age of 3 years but there is great individual variation both in the timing of first words and the level of eventual fluency. Some children can hold simple conversations, others use single words and for others speech is not possible. Other means of communication such as expression, gestures, vocalising and pictures are very important.

Learning

A very few children have little apparent learning difficulty but most need considerable support with their learning. It is hard to predict the extent of difficulties from the chromosomes alone, but observing development allows a clearer picture to emerge.

Behaviour

Features of an autistic spectrum disorder are common in early childhood and may be the symptom that leads to diagnosis. Some children are also very active and easily distractible, and may have a diagnosis of attention deficit hyperactivity disorder. *Unique's* experience shows that some pre-school children have severe temper tantrums. As communication improves, and sometimes with the help of therapy and prescribed medicines, the outbursts usually become much more manageable.

Other concerns

- **Feeding** In the early days, feeding can be hard work for a baby who sucks slowly and around one baby in three with Ring 22 has reflux. This is an extreme form of possetting where milk and stomach secretions flush back up the food pipe. In a young baby, small feeds, careful positioning, feed thickeners and antacid medicines may be all that is needed. For babies and children who have severe or long term reflux, a surgical solution may be considered.
- **Sleep problems** It is worth anticipating sleep problems, because they appear to be unusually common. Firm management may be enough to allow parents to rest but if not, many families also consult specialist sleep clinics and may use prescribed medicines.

Children with Ring 22 are usually healthy. The problems listed below generally only apply to a small number of children.

- **Upper respiratory tract infections** are slightly more common than expected in young children. Middle ear infections are especially frequent, and may lead to repeated insertion of grommets/ventilation tubes into the ear drums. Chest infections and infant wheezing are common in early childhood but are outgrown typically by age 6 to 8, although some children remain asthmatic.
- **Puffy hands and/or feet** are common but not generally troublesome.
- A minority of boys have slightly unusual **genital features** such as undescended testicles, or a hole for urine that is positioned on the underside of the penis (hypospadias). This can usually be corrected in a one or two-stage surgical operation. A small number of children also have a problem with drainage of urine from the bladder.
- **Seizures** A few children develop seizures but anti-epileptic medicines usually keep them well controlled. Seizures can start at any age among both children and adults.
- **Heart anomalies** have been found in a few children. If they do not resolve naturally, they can usually be corrected surgically.