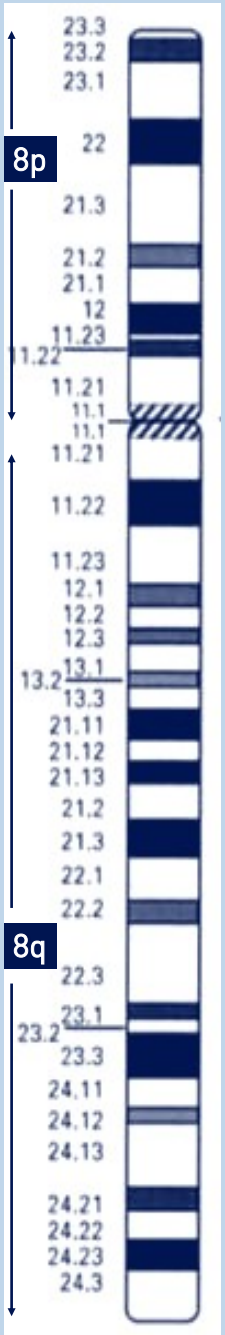
Chromosome 8



Why did this happen?

To answer this question, a geneticist first needs to know about the parents' chromosomes. In some cases, one parent will have a balanced rearrangement of their own chromosomes. The parent is usually healthy and has developed as expected.

In other cases, the parents' chromosomes will be normal and the 8q duplication will turn out to be a sporadic event that has happened out of the blue. The actual cause is then not known but it should be regarded as an event that happened in cell division in the process of making sperm or egg cells. These events 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 duplication to occur.

Can it happen again?

So long as tests show that the parents' chromosomes are normal, they are very unlikely to have another affected child. If the tests show a rearrangement of one parent's chromosomes, there is a greater risk of having another affected pregnancy. Each individual situation is different, and families are recommended to discuss their position with their genetics service.

Inform Network Support

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Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at
www.rarechromo.org/donate Please help us to help you!

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 8q duplications.
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 Maria Cristina Digilio, Medical Genetics Unit, Bambino Gesu Hospital, Rome, Italy and by *Unique*'s Chief Medical Advisor Professor Maj Hulten, Professor of Medical Genetics, University of Warwick 2005.

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Registered in England and Wales
Charity Number 1110661
Company Number 5460413

Understanding Chromosome & Gene Disorders

8q Duplications

rarechromo.org

What is an 8q duplication?

An 8q duplication is a rare genetic condition in which there is an extra copy of some of the material from one of the body's 46 chromosomes, chromosome 8.

Chromosomes are the microscopically small 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. Chromosomes are numbered 1 to 22 according to size and the 23rd pair are the sex chromosomes, two Xs for a girl and an X and a Y for a boy. Each chromosome has a short (p) arm and a long (q) arm.

People with an 8q duplication have an extra copy of some of the material on the long arm of one of their chromosome 8s. Duplication of the whole of the q arm is sometimes called trisomy 8q or partial trisomy 8.

Effects

The precise effects of gaining material from a chromosome vary depending on how large the duplication is, how many genes the duplication contains and what those genes do. The effects may not be limited to the genes within the duplicated piece of chromosome because these genes may interact with other genes on the same chromosome or other chromosomes.

Frequent features

- A degree of developmental delay
- Some degree of learning difficulty
- Anomalies of the structure of the heart. These may be simple or more complex and may need surgical correction
- Problems with the bones and joints, specifically missing or extra ribs, absent or small kneecaps, clenched joints
- Anomalies of the genitals and/or urinary systems. In boys, this may include undescended testicles, a small penis and perhaps a small scrotum
- Facial similarities. These can include large ears, a horizontal crease below the lower lip, a 'pouting' lower lip and a short neck that allows the chin to rest direct on the chest
- Similarities of body build. These can include widely spaced nipples and a long, slender upper body with sloping shoulders

Development

Feeding

Many babies have feeding difficulties as new-borns, often sucking weakly, making breast feeding difficult. Some may be helped by tube feeding (a tube is passed through the nose and into the stomach) or gastrostomy feeding direct to the stomach. Some babies may have acid reflux (where stomach contents flush back up the food pipe) which can be treated with careful feeding, feed thickeners, medication and if necessary surgery. In *Unique*'s experience, by the age of two or three, children are typically eating a variety of family foods and drinking from a spouted cup or bottle.

Mobility and activity

The age at which babies achieve their mobility milestones of holding their head steady, sitting up, scooting, rolling or crawling and walking is likely to be delayed. Hypotonia (low muscle tone) is common in babies and may continue into childhood especially in the upper body and trunk. Physiotherapy helps to develop and strengthen muscles. Children may need support with a frame to achieve independent walking and may also need splints or braces to support their feet and ankles. Despite the early delay, by school age most *Unique* children were mobile and some were running, jumping and climbing.

Learning

The chromosome disorder will affect the speed at which many children learn and set some limits on their eventual achievement. Just how slight or far reaching the effects are will become clear as your child develops, but a moderate learning difficulty is generally believed to be common. It is not always possible to predict from the chromosomes what the effects on learning will be, but by your child's first birthday it will be easier to suggest the outlook from the chromosomes, the results of clinical investigations and observations of your child's development.

Speech and communication

Speech and language are typically delayed and there is some evidence that understanding outstrips expression. Children are likely to benefit from learning sign language and using pictures to stimulate communication before and after words emerge and may well use other means to communicate, including gestures, facial expression and vocal noises.

Medical concerns

The medical concerns listed here are common in a group of individuals with an 8q duplication. Your baby or child may not be affected at all and if they are, to a slight or greater degree.

Heart

Some babies are born with an anomaly of the structure of the heart. In *Unique*'s experience of 35 children, the most frequent heart problem was a small hole between the upper or lower chambers of the heart. This may resolve naturally or need to be closed with surgery.

Skeleton

The development of the skeleton appears to be affected in specific ways: the kneecaps are typically small or missing, the trunk is slender, the shoulders sloping and in some children there may be underdeveloped or extra ribs. Over time, the chest may become hollowed in a formation known as pectus excavatum. In the feet, the heelbone can be prominent, the toes may be crooked and there may be a 'sandal gap' between the first and second toes.

Spine

There is a tendency for the spine to develop a curvature during childhood. At first the degree is often not severe.

Mouth

Many children have either a high roof to the mouth (a high arched palate) or a split in the palate or soft tissue at the back. A cleft interferes with both feeding and speech and can be closed surgically.

Limbs and feet

Many babies are born with their feet held in an abnormal position. This usually needs surgical correction but in some children physiotherapy and bracing may be enough to mould the feet into a good position for walking.

Genital area, reproductive tract and bottom

Boys may be born with undescended testicles and sometimes a small penis or scrotum. In girls, visible anomalies are less common. The hole for the bottom (anus) may be far forward, calling for careful nappy/ diaper hygiene. Structural anomalies of both kidneys and urinary tract appear to be common.

Eyesight

Some children may have a squint (strabismus) and ptosis (hooded upper eyelid) may also occur. If necessary, both conditions can be corrected with surgery.