Why did it happen?

A chromosome 7q deletion can occur as a result of rearrangements in one parent's own chromosomes or it can happen out of the blue.

If an analysis of the parents' chromosomes reveals a structural rearrangement, this is usually balanced so that all the chromosome material is present, and the parent is then almost always healthy. Very occasionally the rearrangement will be the same as in the child, and again the parent may be healthy or they may have similarities with their child.

When the deletion happens out of the blue, the actual cause is not known and it should be regarded as something that happened in cell division during the process of making sperm or egg cells. This is not uncommon and 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 loss of chromosome material.

Can it happen again?

Where both parents have unaffected chromosomes, it is unlikely that another child will be born with a 7q deletion. Where a parent has a rearrangement of their chromosomes, the risk of having another affected child is higher. Where one parent has the same 7q deletion as the child, the risk of passing it on can be as high as 50 per cent.

Inform Network Support

Unique
Understanding Chromosome & Gene Disorders

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Join Unique for family links, information and support. Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website: 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 detailed leaflet about 7q deletions.

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. The content of the full leaflet from which this information sheet was derived was verified by Professor Jean-Pierre Fryns, Center for Human Genetics, Leuven, Belgium and by *Unique*'s chief medical advisor Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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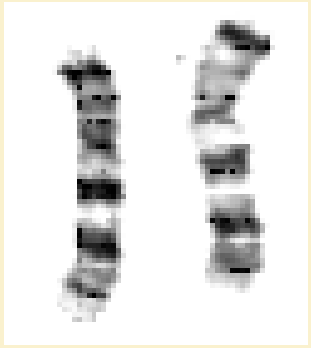
7q36 deletions

rarechromo.org

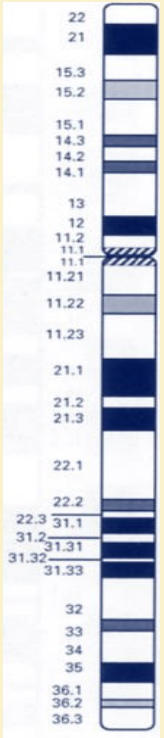
What is a 7q deletion?

A chromosome 7q deletion is a rare genetic condition in which there is a missing copy (deletion) of part of the genetic material that makes up one of the body's 46 chromosomes: chromosome 7.
The genes that contain coded instructions for the body to develop and work as expected are located on chromosomes. Normally there are 46 chromosomes in a cell, occurring as 23 pairs. One of each pair comes from the mother in the egg, the other from the father in the sperm. There are 22 pairs of chromosomes numbered from 1 to 22, roughly from longest to shortest. The 23rd pair are the sex chromosomes. Girls and women have two X chromosomes and boys and men have an X and a Y chromosome. Each chromosome has a short arm ('p') and a long arm ('q').

Chromosome 7



A pair of chromosome 7s as seen under a microscope



A diagram of chromosome 7 (called an ideogram)

Effects

No two people are affected in exactly the same way by a chromosome disorder. A child may show features so mildly that you would hardly notice them or more obviously. Some people have no obvious features of a chromosome disorder at all and most *Unique* members were diagnosed not because of medical problems but because of developmental delay.

The likelihood of developing symptoms and features depends very much on the genetic material that is missing. The following features have been described most often in medical publications and are most likely to make a difference to a child's health or development:

- Some difficulty with learning. This may be borderline or mild or more severe
- Feeding problems
- Floppiness (hypotonia)
- Underdevelopment of the middle of the face
- Underdevelopment affecting the head and sometimes the brain. The head can be very small (microcephaly)
- Malformations of the base of the spine (sacrum)
- Anomalies of the sex organs (genitals) in boys

Genes on 7q

Genes near the end of the long arm of chromosome 7 affect development in specific ways. There are clinical consequences in some people with a 7q deletion but not all.

Holoprosencephaly (HPE). The absence of the Sonic Hedgehog (*SHH*) gene at 7q36 is associated with a developmental disorder called holoprosencephaly. This affects the brain and often the central part of the face. Effects range from scarcely noticeable to severe. A mild form of HPE might show as a single central front tooth, cleft palate or absence of the sense of smell. In its most severe form, the brain fails to develop into two halves (hemispheres).

Sacral defects . The *HLXB9* homeobox gene sited between the *SHH* gene and the tip of the chromosome affects the development of tissues that have their origin in the tail bud of the embryo. The lowest bones of the sacrum and coccyx at the base of the spine may fail to form properly. Other tissues may be involved, including the anus (the hole for the bottom), the rectum, the genital system, the urinary tract and the bladder. Apart from affecting the lower bones in the sacrum, the most common features are chronic constipation and the growth of a cyst, fatty lump or other mass near the sacrum.

Development

Growth

Like many other children with chromosome disorders, some babies with 7q deletions are born small for dates and grow slowly, remaining very short as children and as adults. However, this pattern is not universal.

Learning

The range of effects on the ability to learn is quite varied. Generally, some support with learning will be needed although difficulties range between mild and severe. Very occasionally a very small deletion within 7q36 has been found in people with no learning difficulties.

Speech and communication

Children generally experience some delay in acquiring speech. In general, children seem to show a strong wish to communicate so that in the few cases where speech does not emerge, children use a rich variety of other means to communicate.

Behaviour

Children generally appear to be pleasant, sociable people and this puts them at an advantage at school and later in life in their placements. Some families have noted a strong streak of stubbornness that can develop into challenging behaviour.

Sitting, moving, walking

Some delay in sitting, crawling and walking is very common but the range of eventual mobility is extremely wide. In general children with larger deletions seem to be more delayed, to have muscular floppiness (hypotonia) and to need supports and walking aids. But at least one child with a large deletion from 7q32 is active and sporty. Children in whom the lower spinal area (sacrum) is affected may have specific difficulties with mobility and balance. Loose and easily dislocatable joints are common. Some children with flexible ankle joints have needed orthotic supports but there is improvement with time and few children have needed surgery.