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 normally.

In other cases, the 7q duplication will turn out to be sporadic, that is, it has happened out of the blue and the parents have normal chromosomes.

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 and there is no reason to suggest that your lifestyle or anything that you did caused the chromosome 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.

How rare are 7q duplications?

These are rare chromosome disorders. Only around 20 babies and children with a pure 7q duplication have been described in the published medical literature. It is more common to have a 7q duplication with a duplication or loss of material from another chromosome as well.

At the time of writing, *Unique* had a small community of 12 member families with a pure 7q duplication. Member families who wish to do so can make contact with each other.

Inform Network Support

Understanding chromosome disorders Unique

Rare Chromosome Disorder Support Group,
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Tel/Fax: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

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 duplications of 7q.

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 Steve Scherer, The Hospital for Sick Children, Ontario, Canada and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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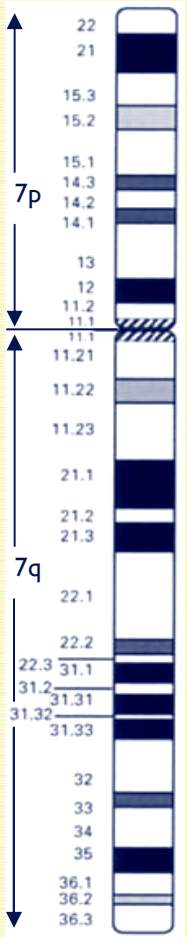
Understanding chromosome disorders Unique

Duplications of 7q

rarechromo.org

What is a 7q duplication?

Chromosome 7



A 7q duplication is a rare genetic condition, in which there is an extra copy, known as a duplication, of part of the genetic material that makes up the body's chromosomes.

Chromosomes are the microscopically small structures in the nucleus of the cells that carry genetic information. They are numbered in pairs from 1 to 22, from largest to smallest, with one member of each pair coming from the father and one from the mother, in addition to the sex chromosomes, X and Y for a boy and two Xs for a girl.

Each chromosome has a short (p) and a long (q) arm. People with a chromosome 7q duplication have a duplication of some of the material from the long arm of one of their chromosome 7s.

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 on other chromosomes.

Main reported effects

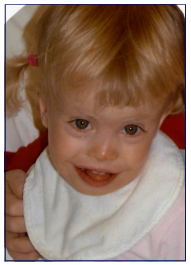
When a chromosome disorder is as rare as a 7q duplication, it is not possible to be certain how it will affect an individual child. Very few babies have been described with a duplication (a trisomy) of the entire long arm of chromosome 7. In babies and children with a smaller 7q duplication, the most common features are also common in children with other chromosome disorders.

- Developmental delay
- Some degree of learning difficulty or disability
- Low muscle tone, so the body feels floppy
- Low birth weight and slow weight gain in babies. Eventual height is often short, although not always for those with a small duplication at the end of the chromosome
- High palate (roof of the mouth). There may be a split (cleft) in the soft or hard part of the palate
- Noticeably large head and, at birth, a very large soft spot (fontanelle) on top
- Most babies are healthy at birth, although there may be a heart condition. Occasionally the structure of the brain is affected
- Unusual position of one or both feet (talipes, club foot)
- Curvature of the spine
- Unusual facial features. Most of these are of no consequence to the child and are usually quite subtle. They might include a high, rounded forehead, downslanting and wide set eyes, skinfolds across the inner corner of the eye, a small, short nose with a low bridge, a small chin or jaw, ears with an unusual shape that are placed low on the head and a short neck.

Duplications of different bands

Small duplications involving 7q21 and 7q22

The most consistent effects are common in youngsters with a rare chromosome disorder: developmental delay; learning difficulties, typically moderate; initial feeding difficulties and failure to thrive; low muscle tone (floppiness) and unusually flexible joints; frequent respiratory infections in childhood; strabismus (squint); short stature, with a relatively large head but no evidence of structural brain abnormalities; some behaviour difficulties and problems with social integration.



Alert and interested: 7q32 duplication

Duplications of different bands

Duplications from 7q21 or 7q22 to 7q31 or 7q32

The main features are all common in children with other chromosome disorders: usually mild to moderate developmental delay; low muscle tone and unusually flexible joints; growth delay; typical facial features including a rounded forehead, small eye openings, widely spaced eyes with skin folds across the inner corners, squint (strabismus) and low set ears; frequent respiratory infections in children.

Duplications from 7q31 to the end of the chromosome

The rate of health problems in the first year of life seems to be high. Common features, most of them frequent in children with other chromosome disorders, include: low birth weight; developmental delay; learning difficulties; growth delay; feeding difficulties; unusual brain structure; heart problems; underdeveloped lungs; cleft palate; unusually large fontanelle (soft spot); skeletal anomalies, for example, missing 12th ribs.

Duplications from 7q32-7q35 to the end of the chromosome

Babies and children with this duplication, often linked with a deletion from another chromosome as part of an unbalanced chromosome translocation, appear to do better than those with a larger duplication extending into band 7q31. The most consistent features are quite non-specific and include a birth weight within the normal range and subsequent normal growth rate, a variable degree of developmental delay and learning difficulty, a relatively large head, some feeding difficulties and some unusual genital features in boys such as undescended testicles.

Duplications from 7q36 to the end of the chromosome

There is little experience with individuals with a small duplication of material near the end of the chromosome, but it appears that effects are less far-reaching than when the duplication is larger. Common features include:

- Some degree of developmental delay, especially speech
- Large head with a prominent forehead.