Why did this happen?
A chromosome 21q deletion can occur as a result of rearrangements in one parent's own chromosomes or it can happen out of the blue, so the child with the chromosome disorder is the only affected person in the family.

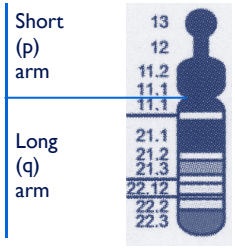
If an analysis of the parents' chromosomes reveals a structural rearrangement in the chromosomes, this is usually balanced so that all the chromosome material is present, and the parent is then almost always healthy. Occasionally the rearrangement will be the same as in the child, and again the parent may be healthy or they may have similarities with their child. In this case, the deletion does not usually include band 21q22.

How did this happen?
Rearrangements occur in chromosomes as part of evolution. 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 loss of chromosome material. Changes to the structure of chromosomes such as 21q deletions occur most often during the cell divisions that lead to the creation of eggs or sperm.

Can it happen again?
Where both parents have normal chromosomes, it is unlikely that another child will be born with a 21q 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 21q deletion as the child, the risk of passing it on can be as high as 50 per cent.

How rare are 21q deletions?
They are almost certainly very rare, but an exact figure cannot be given because some people are not affected by their chromosome loss or are so mildly affected that they do not come to medical attention.

Chromosome 21



Inform Network Support

Understanding chromosome disorders Unique

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When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a full leaflet about the effects of 21q 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 and the text of the full leaflet on which this short version is based was verified by Dr Julie Korenberg, Cedars-Sinai Medical Center, Los Angeles, California, US and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK, 2005.

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Understanding chromosome disorders Unique

21q deletions

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What are 21q deletions?

A chromosome 21q deletion is a rare genetic condition in which there is a missing copy of part of the genetic material that makes up one of the body's 46 chromosomes. Chromosomes are the microscopically small structures in the nucleus of the body's cells that carry genetic information. They come in different sizes, each with a short (p) and a long (q) arm. Chromosome 21 is the smallest chromosome, containing no more than 200 to 400 genes. The likelihood of problems developing depends very much on what genetic material is missing.

The short (p) arm of chromosome 21 is very small and similar genes are available on other chromosomes, so it can be lost without apparent harm. Losing material from the long (q) arm is more important, particularly when material is lost from the bottom bands at 21q22. However, losing the final tip of 21q22.3 is also thought to be harmless. A geneticist or genetic counsellor can tell you exactly how much material has been lost and where the breakpoints are in the chromosome.

Main features

From band 21pter to band 21q21

Many people with material lost between the tip of the short arm and upto and including the very first part of band 21q21 are healthy, have no learning difficulties or need only a small amount of support and go on to lead independent adult lives. Some people with more material lost from the region of 21q21 do experience difficulties with learning. They may also have birth defects, but the pattern is very variable and from what is known today unfortunately cannot be predicted from the amount of chromosome material lost.

Including band 21q22

- A variable degree of learning difficulty is fairly common.
- At birth, the baby's head may be very small (microcephaly) and it may grow slowly.
- Joints may be stiff and held in an unusual position.
- The muscles of the body may feel either too tight (hypertonia) or too floppy (hypotonia).
- A few babies may be born with a cleft lip or a cleft palate (a split in the roof of the mouth). This is repaired with surgery and meanwhile help is given with feeding.

Development in affected children

Food and eating

Babies and children who have deletions that do not include band 21q22 generally feed well from birth. Among babies and children with a deletion including band 21q22, difficulties are more common and families usually benefit from feeding support, particularly as many babies do have great difficulty in putting on weight.

Learning and schooling

In many instances people with a deletion that ends within band q21 or at q22.1 have not apparently had any learning difficulties. Others have had slight learning difficulties, particularly in secondary education. A smaller number have had a moderate learning difficulty.

For people with a deletion that includes bands 21q22.1 to q22.3, more difficulties with learning can be anticipated and children usually benefit from special education.

Communicating and speaking

Generally, children have a delay in speech and language skills that is in line with their level of learning difficulty. As a broad generalisation, children in whom 21q22 is preserved intact are more likely to acquire speech and to use it to communicate their needs and wishes than children with a deletion involving 21q22.

Sitting, walking, moving

In general, children are more likely to be late in sitting, walking and running than children without a chromosome disorder but some are quite mildly affected.

Underlying the delay is an alteration in muscle tone, so that the muscles either feel too tight and tense (hypertonia) or too floppy and loose (hypotonia). Most children have a mixture of hypertonia and hypotonia. Physiotherapy is helpful to achieve and maintain mobility.

Behaviour

Children with speech and language difficulties may have behaviour problems due to frustration. Additionally, some children with 21q deletions have a short attention span and some hyperactive behaviours. Children with behavioural difficulties should be evaluated by a psychologist, psychiatrist or developmental paediatrician who can help provide guidance, intervention strategies and medications if needed.

Two years old, with a 21q22.1q22.3 deletion

Medical concerns

Respiratory infections

Respiratory infections appear to be common in babies and young children. Your child's medical carers will indicate sensible precautions, including full immunisation.

Joints

A baby born with one or more joints held in a bent position and unable to be fully flexed can be treated with physiotherapy (physical therapy), splinting and casting to stretch the joints and to hold them in place, and if needed with surgery.

Blood disorders

A mild degree of thrombocytopenia can occur, where levels of the platelets that help to form blood clots are low. In this case, cuts and nosebleeds would take longer to clot and bruises could be extensive.

Heart conditions

Babies will have a thorough heart exam and a detailed cardiac evaluation if abnormal heart sounds are heard. Examples of heart abnormalities that have been seen include holes between the upper and lower heart chambers (atrial or ventricular septal defects, ASDs, VSDs), narrowing of the blood vessel that takes blood to the lungs to pick up oxygen (pulmonary stenosis) and underdevelopment of certain heart valves. In some cases the abnormality resolves in time, but surgery may be needed. Babies generally thrive after surgery.

Brain

A gene situated at 21q22.3 is involved in the normal development of the brain. Disruption of this gene can cause problems which may be mild (for example, the glands for smell are missing) or more obvious.