

How common are deletions spanning 5q22?

This is not known, but they are very rare. At the time of writing, *Unique* had 17 member families with this deletion.

Why does my child have a chromosome disorder?

Most often, a chromosome 5q deletion happens out of the blue, and the affected child is the only person in the family with a chromosome disorder. Geneticists call this type of event *de novo*.

Occasionally the deletion can occur as a result of a rearrangement in the chromosomes of one of the parents. The only way to know is for the parents' chromosomes to be checked.

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 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 normal chromosomes, it is very unlikely that another child will be born with a 5q deletion. Where one parent has an atypical chromosome rearrangement, the risk of having another affected pregnancy is much higher. A clinical geneticist can give you specific advice for your family.

Families say ...

Loving and lovable and has an individual sense of humour - age 13.

She causes me to look at things more simply. She knows what's really important - age 11.

Her smile lights up a room - age 5.

A learning experience for me every day. Patience, love, understanding - age 4.



For support,
contact with other families and information

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Support Group**

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When you are ready for more information, *Unique* can help. We can answer individual queries and we also publish a longer leaflet about the effects of 5q deletions.

This information sheet is not a substitute for personal medical advice.

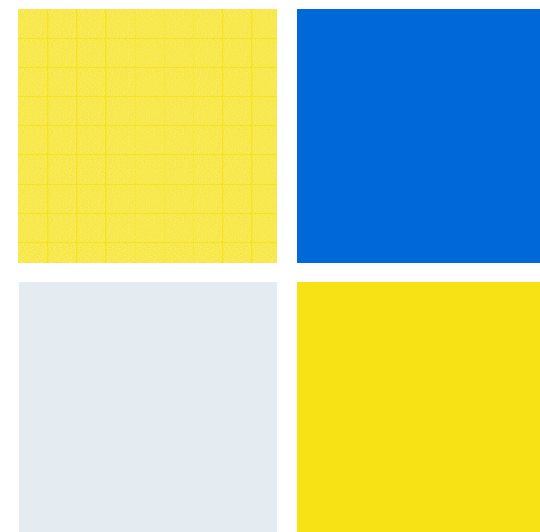
Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the medical content has been verified by

Dr Diana Eccles, Wessex Regional Genetics Service, 2004
and Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

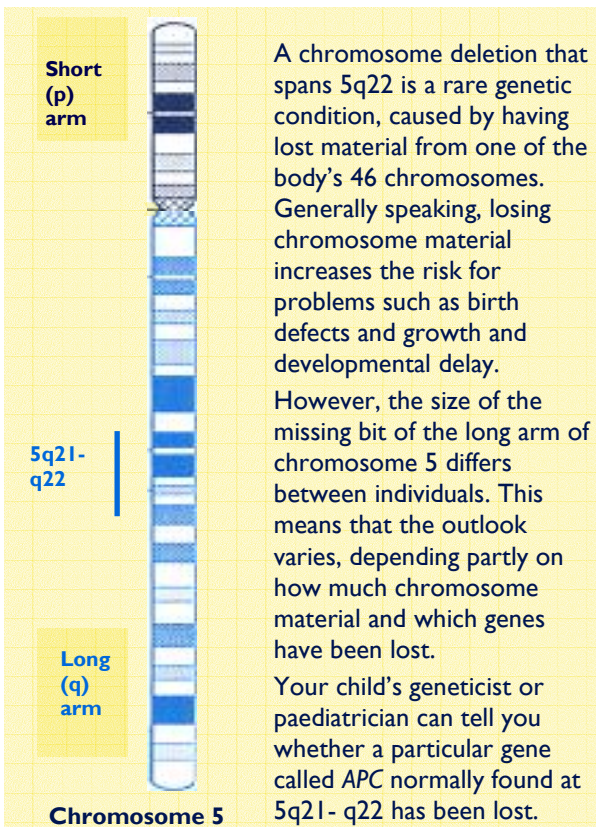
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Deletions including 5q22



What is a deletion spanning 5q22?



A chromosome deletion that spans 5q22 is a rare genetic condition, caused by having lost material from one of the body's 46 chromosomes. Generally speaking, losing chromosome material increases the risk for problems such as birth defects and growth and developmental delay.

However, the size of the missing bit of the long arm of chromosome 5 differs between individuals. This means that the outlook varies, depending partly on how much chromosome material and which genes have been lost.

Your child's geneticist or paediatrician can tell you whether a particular gene called *APC* normally found at 5q21-q22 has been lost.

APC gene

Your child's geneticist or paediatrician can tell you whether a particular gene called *APC* (adenomatous polyposis coli) normally found at 5q21-q22 has been lost. This is because people who have lost one copy of this gene have a higher than normal chance of developing a type of colon cancer called FAP (familial adenomatous polyposis) as young adults. If so, your child will normally be screened from the age of 10-12 years, or younger if they have any bowel problems or bleeding.

You can find out more about FAP by reading the *FAP* leaflet on the website of the British Society for Human Genetics at www.bshg.org.uk in Leaflets.

Development

■ **Size, growth and feeding**

Some babies are born small for dates, others are around their expected birth weight. Almost all babies seem to have difficulty putting on weight and sometimes young children do too. Quite a few children are unusually short, but this is not always the case. Most babies are slow or reluctant feeders and those who have a high arched or cleft palate (split in the roof of the mouth) may need special teats and feeding techniques as well as any treatment that is indicated. Older children may continue to find chewing difficult but in *Unique's* experience weight gain problems ease in childhood.

■ **Sitting, moving, walking**

Almost all children in *Unique* have been late to sit and walk but most eventually walk and a few cycle and swim. This is not possible for all children and a small number remain in a wheelchair. Many babies are floppy (hypotonic) and benefit from physiotherapy to increase their muscle tone.

■ **Ability to learn**

Most children have some learning difficulty but this may be specific and mild; some children attend a mainstream school with support. Other children are more affected and thrive better in special schooling. *Unique's* experience is that the level of learning difficulty is very varied and is easier to predict from watching your child develop than from knowing the exact chromosome constitution.

■ **Speech**

Most *Unique* members use their first words between 3 and 5 years and a few go on to speak in long, complex sentences. Difficulties with pronunciation can be quite persistent so speech may remain unclear. A few children do not speak and communicate using signing and communication technologies.

■ **Behaviour**

Most *Unique* children are loving and warm. They may be vulnerable to insecurity outside their home and familiar surroundings and need careful preparation for changes. Some older children display social immaturity and benefit from social skills teaching.

Medical concerns

These are complaints found in a group of children with a chromosome deletion covering 5q22. An individual child may or may not be affected.

■ **Hips and feet**

Some babies are born with loose hip joints that need stabilising with splints and sometimes with plaster casts. Unusually angled feet are also common and may need treatment to improve walking.

■ **Kidneys and urinary system**

The kidneys and urinary system may be affected. In *Unique's* experience narrowing of the tubes that lead to or from the bladder is common, leading to potential drainage problems and the kidneys themselves may not be normally formed.

■ **Heart conditions**

Any heart conditions appear to be fairly minor and most either resolve naturally in babies or do not need treatment. The conditions noted most often are holes between the chambers of the heart and an unusually formed valve between the heart and the aorta that takes the blood to the body.

■ **Infections**

Children with chromosome disorders tend to catch more respiratory infections than other children and to suffer worse when they develop them.

■ **Seizures**

Some children will have seizures but in *Unique's* experience they will either outgrow them or the seizures can be well controlled with medication.

■ **Hearing**

Unique's experience is that both permanent and temporary hearing impairment is quite common, although the level is variable.

■ **Eyesight**

Strabismus (squint) is the most common and obvious concern but is normally treatable with patching and if necessary surgery. A small number of children appear to be very short-sighted.

Other conditions *Unique's* full leaflet on **Deletions including 5q22** lists other medical conditions that have been found occasionally.