6q deletions
6q11 to 6q16
Proximal deletions of 6q: from 6q11 to 6q16

A chromosome 6q deletion is a rare disorder in which some of the genetic material that makes up one of the body’s 46 chromosomes is missing. Like most other chromosome disorders, this increases the risk of birth defects, developmental delay and learning difficulties. However, the problems that can develop depend very much on what genetic material is missing.

Chromosomes are the structures in the nucleus of the body’s cells that carry the genetic information that controls development and function. In total each one of us normally has 46 chromosomes. Of these, two are a pair of sex chromosomes, XX (a pair of X chromosomes) in females and XY (one X chromosome and one Y chromosome) in males. The remaining 44 chromosomes are grouped in 22 pairs, numbered 1 to 22 approximately from largest to smallest.

One chromosome from each pair is inherited from the mother while the other one is inherited from the father. Each chromosome has a short arm (called p) and a long arm (called q).

You can’t see chromosomes with a naked eye, but if you stain them and use a microscope to magnify their image about 1000 times, you can see that each one has a distinctive pattern of light and dark bands. The image below shows pairs of chromosome 6 from several cells. The right hand chromosome in each pair has a 6q11q16 deletion.

In the diagram of chromosome 6 on the right, the bands are numbered outwards starting from the point where the short and long arms meet (the centromere). A low number, as in q11 in the long arm, is close to the centromere. Regions closer to the centromere are called proximal. A higher number, as in q27, is closer to the end of the chromosome. Regions closer to the end of the chromosome are called distal. When two breakpoints have rejoined, leaving a segment out, the deletion is called interstitial.

The red bar marks the region of the long arm considered in this guide.
Your geneticist or genetic counsellor can tell you more about how much material has been lost in your child’s case. You will be given the results of the genetic test, showing the breakpoints in chromosome 6. Comparing your child with others will help to create a picture of what to expect. But there are often differences, sometimes quite marked, between children with apparently similar test results. It is important to see your child as an individual and not to make direct comparisons with others. Each of us is unique.

Pregnancy
In nine cases where pregnancy was described, it was considered normal in six cases. In one pregnancy where the baby had a 6q13q15 deletion, the baby was very small for dates and had a kidney problem; there was a large quantity of amniotic fluid (polyhydramnios) and after labour started at 30 weeks, the baby was born at 33 weeks. Growth delay and excess amniotic fluid affected a pregnancy where the baby had a 6q14.2q16.2 deletion; the baby was born at 38 weeks. Two mothers of babies with a 6q14q16 deletion noticed much slighter fetal movements than in their previous pregnancies. When one of the babies was induced at 42 weeks, his birth weight was a healthy 7lb 12oz (3.515kg), but 25oz (700g) less than an older unaffected brother. The other baby looked small despite being 11 days beyond term.

At birth
Birth weight at or near term: 4lb 1oz/1.84 kg - 8lb 5oz/3.78 kg

<table>
<thead>
<tr>
<th>What was unusual?</th>
<th>How many affected?</th>
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<tbody>
<tr>
<td>Low muscle tone (floppiness)</td>
<td>Most</td>
</tr>
<tr>
<td>Reluctant or unable to feed</td>
<td>Many, possibly most</td>
</tr>
<tr>
<td>Needed support with breathing</td>
<td>A small number</td>
</tr>
<tr>
<td>Umbilical hernia</td>
<td>11/29</td>
</tr>
<tr>
<td>Sacral dimple</td>
<td>7/29</td>
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<tr>
<td>Tethered cord / other spinal defect</td>
<td>4/29</td>
</tr>
<tr>
<td>Minor anomalies of genitals</td>
<td>6/13 boys</td>
</tr>
<tr>
<td>Cleft palate (split in roof of mouth)</td>
<td>5/29</td>
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Care
Some babies needed immediate care, including one with a cleft palate and small, receding jaw and others needing resuscitation due to respiratory distress. One was hard to feed and could not maintain her temperature. Not all babies needed support at birth: a Unique baby with a 6q11q15 deletion was born a healthy weight with no unusual features and a chromosome problem was only found after she showed developmental delay.

Key references
The first-named author and publication date are given to allow you to look for the abstracts or original articles on the internet in PubMed. If you wish, you can obtain abstracts and articles from Unique.

Some twenty-nine cases with a pure deletion in this area are described, at least eighteen in the medical literature and 10 members of Unique. The oldest member of Unique was 16 years old when this guide was written and the oldest person described in the medical literature was 28 years old. (Grati 2005; Myers 2005; Yu 2005; Hopkin 1997; Kumar 1997; Gershoni-Baruch 1996; Romie 1996; Di Lernia 1994; Roland 1993; Rose 1992; Valtat 1992; Lonardo 1988; Slater 1988; Turleau 1988; Yamamoto 1986; Cerrillo 1985; Young 1985; McNeal 1977; Unique).
Low muscle tone

An unusually low muscle tone, so that the baby feels floppy to handle. Babies with hypotonia tend to lie with their arms and legs loosely outstretched instead of bent at the knee or elbow. When held under the arms, their bodies easily slip through the hands. Low tone may improve with maturity but babies generally benefit from physiotherapy to help them reach their developmental milestones.

“She has low muscle tone all over her body and hypermobility in her ankles, knees and fingers. She has been wearing splints up to her knees since she was two years old. She used to wear a lycra body suit to give her more support. Her spine curved between her shoulder blades. It was postural due to low tone and the lycra body suit helped to keep her straightened.” - 5 years

Feeding

Many babies, perhaps most, had difficulty establishing feeding. They might show no interest in feeding, be unable to suck effectively or to coordinate the actions of sucking with swallowing. In some babies the problems were mild and temporary and it was possible to breastfeed for some months. More commonly, babies needed long-term support and were fed either through a nasogastric tube or a gastrostomy tube (PEG, button) direct into the stomach.

Gastro oesophageal reflux (GORD, GERD), where the stomach contents return up the food passage, affected some babies. Reflux raises a baby’s risk of inhaling feed contents and setting up an infection in the lungs known as aspiration pneumonia. Reflux can be eased by careful semi-upright positioning during and after feeds, sleeping in a prescribed sleep chair rather than a bed, raising the head end of the baby’s cot and if necessary by prescribed medication that helps to keep the feed within the stomach and counteract any acidity. Babies who have continuing problems can have a surgical procedure called a fundoplication to improve the action of the valve at the junction of the food passage and stomach. Where feeding and reflux problems are persistent, a gastrostomy tube can be inserted to allow direct feeding into the stomach until the baby is sufficiently mature to manage feeding by mouth.

“I tried to breastfeed her when she was born but was unable to, as her blood sugar level was dropping. We thought she just wasn’t interested in feeding and she was always below weight. She had a video fluoroscopy at about 2½ years as she had continuous chest infections and was always choking on food and drinks. It showed she had poor coordination of her tongue, a slow swallow reflex and she was at high risk for aspirating liquids. From then on her drinks were thickened and still are now. Her food had to be blended until she was 3 years old. The speech therapist did a lot of work. We had to do facial exercises before meals to stimulate the muscles, then to move her from blended to mashed food we divided her food into three different
bears of different consistencies. We then gave her melt-in-the-mouth type crisps like wotsits, holding them to the side of her mouth to help her hear the sound of biting. Today she really loves food. Her dinners have to be cut up and mashed and food like sandwiches I break into small pieces. She eats well in a high chair and is sometimes able to hold her spoon and get food in her mouth, but I always have a spoon to feed her most of her dinner. She still uses her hands to eat and enjoys them being messy. She always needs supervision when eating as she overfills her mouth. She still needs thickened drinks in a toddler cup with a lid

A small number of babies needed considerable support with their breathing, in part because the wind pipe (trachea) was unusually soft and floppy. In two babies a connection was found between the wind pipe and the food passage (tracheo oesophageal fistula). Three babies were fitted with a tracheostomy to allow air and oxygen to reach the lungs directly and one child still had her ‘trach tube’ in place at the age of six.

Babies with a chromosome disorder are more likely than typically developing babies to have one or more birth defects.

An umbilical hernia is often seen in babies with a proximal 6q deletion and all three babies with a deletion that included band 6q11 had one. This shows as a soft, skin-covered bulge at the umbilicus (navel, belly button) that can look bigger when a baby strains or cries. The bulge contains a small piece of abdominal lining and sometimes a part of the abdominal organs. It is caused by incomplete closure of the ring of muscle that the umbilical cord passed through during fetal life. The hernia may be quite small and can be left to resolve naturally by the age of 3 or 4 years. Some babies have a very large hernia (2”/5cm) or one that does not improve, in which case it can be surgically stitched in a small operation.

A sacral dimple (dimple or hole in the skin just above the crease between the buttocks) is also sometimes seen. This may be shallow so you can see the base, but stools can collect there before your child is toilet trained, so keeping it clean and protected is important. A sacral pit may be deep and even connect to the spinal canal or the colon. If there is any concern about this, your baby’s spine will be imaged, usually with ultrasound or an MRI scan.

The bottom end of the spinal cord is usually free within the spinal column but occasionally it becomes attached to one of the surrounding structures. This is called a tethered cord. A tethered cord can be put under tension as a child grows and moves and this can cause damage to the muscles and nerves that control the legs, feet, bowel and bladder. An MRI image will give a detailed 3-dimensional picture of the spine and
spinal column. If necessary the cord can be surgically released so that it can hang freely. This spinal defect was seen in two Unique children. A third child was born with spina bifida, itself a common cause of tethered cord. One child had an additional vertebra and a 13th pair of ribs, which would not need treatment.

Minor anomalies of the genitals and the bottom area are often seen in babies with a chromosome disorder, especially boys. The most common problem was undescended testicles. The testes descend during fetal life from a position just below the kidneys at the back of the abdomen to reach the scrotum, usually before birth. If one or both testicles remain undescended, a decision will be taken whether to bring them down surgically and anchor them in the scrotum. One boy had a very small penis, another had hypospadias, where the hole for the urethra that is usually at the end of the penis is situated on the underside instead. Hypospadias can be repaired by surgery, the operation usually carried out during babyhood.

Defects in the roof of the mouth (palate) are common in children with and without a chromosome disorder. The hard palate at the front of the mouth may be split or the split may be found further back in the soft, fleshy tissue at the back of the top of the mouth. Occasionally the split is only seen in the tissue that hangs down above the tongue at the very back of the mouth (uvula, known as a bifid uvula when it is split). Babies who have a cleft palate in combination with a lower jaw that is unusually small (micrognathia) and set back from the upper jaw (retrognathia) have what is known as the Pierre Robin sequence.

The Pierre Robin sequence is most likely to cause difficulties with breathing and feeding in babies. If your baby is affected in this way, the paediatrician and nurses will advise you how to minimise problems. Occasionally a surgical procedure may be needed to ease breathing and the jaw may be extended using a surgical procedure called a jaw distraction. A cleft palate causes difficulties both in feeding and in speech production. Surgical repair of the palate eases these difficulties and may eliminate them altogether.

**Appearance**

There may be little sign in the facial appearance of most babies with a proximal 6q deletion of the underlying disorder. Doctors and sometimes parents may notice what are known as ‘dysmorphic features’ but these are usually of little or no importance to the baby. Children will not have all of these features but they may have some and they may look rather like other children with a proximal 6q deletion.

Some features are seen in many babies and children with a chromosome disorder, others are more specific to a proximal 6q deletion. The features typical of many chromosome disorders include ears that are set below the usual level (in line with the
eye) and may be large, floppy or oddly formed, a remarkably small lower jaw and chin (micrognathia), eyes that slant slightly upwards, tiny skin folds across the inner corner of the eye (epicanthic folds), a short neck, eyes set wide apart and a flat bridge to the nose.

Features more specific to babies or children with a proximal 6q deletion include a thin upper lip, an asymmetric face, a short, sometimes upturned nose with a bulbous tip, a long or large groove between the nose and upper lip, rounded, chubby cheeks and some loose skin around the neck or more generally on the body.

"The first different thing I noticed was her eyes. My other children have round, wide eyes but hers were smaller and slanted. The bridge of her nose is flat, she has a small, slightly receding chin, a short neck, large ear lobes and small, slanting shoulders. But to look at her you wouldn’t know anything was wrong, as she is a very pretty girl - 5 years"

**Hands and feet**

Minor anomalies of the hands and feet are relatively common in children with chromosome disorders. These may just be cosmetic or they may make it harder for the child to use their hands or to walk. Hands may be short, although in a number of cases they are long and slender and the fingers either unusually flexible or clenched. Clenched fingers can sometimes be straightened with regular splinting. The fifth finger may curve in towards the hand. The thumb may be set unusually low.

Some babies have unusually small feet and there may be webbing between the toes (most often the 2nd and 3rd toes). A prominent heel can be typical, as are very flat feet or a sole that forms a curve like the rocker on a chair. Two babies were born with feet held in an odd position that would need correction by stretching, physiotherapy, splinting or surgery and casting to help them to walk properly.

"She has lax joints in her fingers and she holds her fingers up and bent a bit. Her knees bend in, again due to lax joints, and her feet are completely flat so that the bone on her inner feet becomes red and swollen due to the pressure" - 5 years
Growing
Babies and children with a proximal 6q deletion can be tall, medium or short for their age, but most commonly they are short, and their height and weight may put them in lowest three per cent of the child population for height and weight. Children who are persistently very short may be referred to a growth clinic and treatment with growth hormone considered.

Medical concerns

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**Head and brain**
A baby’s or child’s head may be unusually small (microcephaly), normal or even large (macrocephaly). Some children have an unusually shaped head but this is not usually important. Where there is any concern, the brain may be imaged to assess its structure and growth and in a few babies and children the fluid filled spaces within the brain (ventricles) have appeared enlarged or asymmetrical in size. In two children the band of nerve fibres that connects the two hemispheres of the brain (corpus callosum) has not been complete. What these findings may mean for an individual child is not always clear but your child’s doctors will explain them. One child had a structurally normal brain which showed however that the natural process of insulation of the nerves known as myelination was slightly delayed.

**Heart**
Defects in the structure of the heart are seen quite commonly in babies with a chromosome disorder. In babies with a proximal 6q deletion the defects resolved naturally in time or were relatively minor and from what we know, those children who needed surgery thrived afterwards. The most common problems were a hole between the two lower (pumping) chambers of the heart (ventricular septal defect/ VSD) or the upper storage chambers (atrial septal defect/ASD) and an open channel linking two major blood vessels leaving the heart that normally closes after birth (persistent ductus arteriosus/PDA). Dextrocardia, where the heart is sited to the right rather than the left, was also seen but this does not need treatment.

**Kidneys**
Six babies had one of a variety of kidney problems, including the kidneys being sited in an unusual position and a missing kidney. It is likely that your child will have a thorough examination of their kidneys and the drainage system for urine, and this is especially likely if they have a urinary tract infection. One problem can be vesico-ureteric reflux,
where urine can flush back from the bladder and potentially damage the kidneys. First-line treatment is usually to give low-dose long-term antibiotics to prevent urinary tract infections. Some children will outgrow the tendency to this type of reflux but others may need a surgical operation to correct the problem causing the reflux. In this operation, called ureteric reimplantation, the ureters (the tubes that conduct urine from the kidneys to the bladder) are disconnected from the bladder and re-attached at an angle to create a valve.

- **Inguinal hernias**
  Three babies or children each with a deletion between 6q13/14 and 6q15/16 had an inguinal hernia. This shows as a bulge in the area where the lower abdomen meets the upper thigh (the groin). The cause is that an opening in the lower part of the wall of the abdomen that is open during fetal life but closes before birth does not in fact close. The remaining opening may be small, only allowing fluid through, or it may be large enough for something such as a loop of the intestine or another organ to get stuck in it. An inguinal hernia should always be assessed by your child’s doctors and your child may need surgery to repair it.

- **Eyesight**
  Twelve babies or children had strabismus (a squint). This can affect one eye or both and the direction may be inward or outward. Severity also varies, with the condition resolving naturally in some babies, but requiring surgical correction in others. Nystagmus, making the eyes move constantly, was seen in four children. It was particularly noticeable when the child was tired. Nystagmus can be related to vision or caused by an imbalance in the muscles controlling eye movement. Your child will have a detailed eye examination to establish the cause and start any treatment needed.

  “She has depth perception problems which was a big issue to her, but the older she is getting she is learning to cope with her surroundings” - 5 years

- **Hearing**
  Three children have had the fluctuating temporary hearing impairment caused by fluid within the middle ear (glue ear). This is a common condition in young children whether they have a chromosome disorder or not. It may resolve naturally in time but can be relieved if necessary by placing aeration tubes in the eardrum. One child was described as hypersensitive to noise.
Teeth
Dental problems are fairly common in children with chromosome disorders who generally require specialist dental care. The underlying development of the structures of the face may be abnormal, affecting the development of the teeth; differences in feeding and mouthing experiences also impact on tooth development. Among children with a proximal 6q deletion, one child had two teeth fused into one, a baby was born with visible teeth and two children had missing teeth, in one case the upper lateral incisors.

She has good teeth. We found it difficult to brush her teeth but have found she will tolerate an electric toothbrush. She drools excessively and her ENT consultant has suggested an operation to move the saliva glands when she is older. In the meantime we will consider botulinum injections. We tried hyoscine patches but they broke her skin down” - 5 years

General wellbeing
The evidence from Unique is that once babies have overcome initial problems and a vulnerability to coughs, colds and common respiratory infections such as bronchiolitis in early childhood, they are generally as healthy as other children. However, the outlook for an individual child is determined largely by their clinical problems. Two babies in this group are known to have died, each in the first month of life. One baby died shortly after being born prematurely at 33 weeks; another shortly after a tracheostomy tube was fitted to enable him to receive air and oxygen.

Channelopathy One girl with a 6q14.1q16.1 deletion had episodes of one-sided weakness, usually after activity, starting around age 4. These were eventually diagnosed as a channelopathy neurological disorder and treated with flunarizine, a drug used for migraine (Unique). A channelopathy is a disease caused by a defect in cell proteins called ion channels causing defects in the movement of materials in and out of cells. The most likely genes underlying these episodes are two called GABRR1 and GABRR2, which are to be found in band 6q15.

Sitting, moving: gross motor skills
Babies and children with a proximal 6q deletion typically face some delay in reaching their mobility milestones but the extent of the delay is extremely varied. From rolling over between seven and 18 months and sitting between four months and two years, some babies progress to crawl (one to three years), while others scoot or bottom shuffle (18 months to four years). First steps are taken between 18 months and six years and independent walking has been achieved between 18 months and 21 years. Most children have considerable hypotonia (low muscle tone, so they feel floppy) and loose, very mobile joints and need support (splints, braces, walkers) to enable them to stand and walk at first. One child also had easily dislocatable hips requiring surgery before he could walk. Another child needed the tendons in the ankles lengthened to achieve a proper heel strike on the ground.

“His gross motor skills are developing well; he is both walking quickly and climbing but is not running or jumping yet. He remains unstable and falls frequently but this has improved. He has been discharged from physiotherapy” - 3 years
“On the go most of the time, he does not like to sit down” - 3 years

“She wears splints on her feet that come up to her knees. She can climb stairs now with help and come down on her bum but when she’s tired she can’t even climb one step. She enjoys swimming but can’t cope with a lot of physical activity as she tires easily. People make the mistake of Thinking she never stops and they are right but it’s not because she has the energy: she is sometimes beyond exhaustion but can’t switch off and won’t rest. Her low muscle tone drops and you can feel the difference in her weight when this happens” - 5 years

**Using their hands: fine motor and coordination skills**

Hand and eye coordination skills such as holding a bottle and playing with small toys may not develop in line with gross motor skills. Overall, there appears to be fairly consistent delay in hand use and fine motor skills. This delay means that early intervention by occupational therapy to stimulate hand use is vital.

“Delayed pincer grip; she has no patience with forks and uses spoons and her hands when eating. She can put on a hat but needs help dressing” - 3 years

“He is still awkward due to his lax ligaments and very flexible hands and fingers. He can use a spoon and put a toothbrush in his mouth and chew it but he needs help with all the activities of daily living” - 3 years

“She has severe developmental delay, delayed gross and fine motor skills, no pincer grip and holds a pencil with all of her hand around it” - 5 years

**Speech and communication**

Some delay in the emergence of speech and language is to be expected, but the extent of the delay is very variable and probably reflects the level of cognitive ability. Some two-year-olds are talking in sentences, others communicating by expression, gesture and vocal noises. Understanding is usually more advanced than expression, with at least one child believed to understand what any other child might. Some young children make good progress when they are taught to sign their needs. It is not possible to say whether all youngsters with a proximal 6q deletion will eventually acquire some speech but it is clear that some will talk meaningfully in sentences.

Despite an inability to form words, some young children with a chromosome disorder are able to sing or hum in tune.

“She attempts singing and can hit the same notes as other people” - 3 years

“Although he does not speak much, he loves singing (humming) nursery rhymes and TV theme tunes. We often hear him singing in the morning” - 3 years
“Speech is her strongest area. She can put 5-6 words together and most of her words are clear. People think she knows everything because they hear her saying so many words but when she was assessed recently they said she does not have the understanding to back up her speech: she echoes what she hears a lot. Her understanding is at a 2-word level. She is very vocal and she can let you know what she wants but she is unable to answer a question at all” - 5 years

**Learning**

A child with a proximal 6q deletion can be expected to have some learning difficulties or disabilities but it is not possible to predict the level of difficulty just from the karyotype. It appears that most children have a mild to moderate learning difficulty but in some cases it may be severe. Some children face particular difficulties with concentration and attention. Children with hypermobile joints and low muscle tone are likely to find holding a pencil difficult. Among strengths identified by families are a facility for music, a wish to identify with other typically-learning children and a determination to learn. One child with a 6q13q15 deletion had particularly advanced visual motor problem solving skills and at pre-school age only a borderline to mild delay in development.

Formal academic skills may never be the focus of the learning experience but there is evidence that at least some children are reading, identifying colours and using number by the age of seven or eight.

“Her memory is not bad although she does forget things like matching and we have to start over again. She can learn music more easily - 5 years

**Behaviour**

The evidence from Unique is that most children have a warm, sociable nature. Like any other child, they can feel frustrated and behave destructively or be overwhelmed and lose control of their emotions.

“She is happy and laughs and smiles a lot; she runs ecstatically towards us and hugs us when picked up from daycare. She most enjoys toys with sound, the piano, people singing and songs with clapping or signs and gestures. She can play with animals for hours. When she feels misunderstood (we believe) she can bite, hit and pull hair.” - 3 years

“Warm, caring nature”

“He is generally good humoured unless thwarted but can become very upset if not getting to do what he wants” - 3 years

“She slept her first year away and at 22 months was still a very tired baby requiring more sleep than the average child of her age. She has sensory processing dysfunction and likes to chew everything and eat things like washing powder or play dough, so we have locks on all cupboards. She was diagnosed with autism when she was 4 and as a result of her communication problems, sensory processing dysfunction and autism she has severe behaviour problems 7 days a week. I can’t express how challenging it is. She has thankfully stopped head butting, she used to have black eyes and swollen bumps on her head. Now her face is permanently covered in scratches were she pulls
her face and rips her hair out. She bites her hands. She also hurts all family members and close friends by biting, kicking, pulling our hair. We are very limited to where we can go outside of the house. Her behaviour is the worst part of her syndrome.

The ironic thing is she is well behaved at school or when she has someone 1:1 working with her giving her lots of stimulation and everything is on her terms. She hates her routine being broken. She interacts well on her terms: there are days when she can’t cope with noise and people and wants to sit in front of the TV on her own with her programmes on over and over again. She becomes distressed some days and bangs her ears if people are too noisy. She is gentle with other children but is aggressive with her sisters - 5 years

“She enjoys listening to music, swinging, chewing toys, watching videos. She is happy mostly” - 6 years
How did the chromosome deletion occur?

A proximal 6q deletion usually occurs out of the blue as a sporadic event when both parents have normal chromosomes. The genetic term for this is *de novo* (dn). More rarely, it can be inherited as a result of a rearrangement in one parent’s chromosomes. A blood test to study the parents’ chromosomes will determine which of these two alternatives occurred in your case.

If the blood test reveals a structural rearrangement in one of the parents, it is usually balanced so that all the chromosome material is present, and the parent is then almost always healthy.

*De novo* 6q deletions are caused by a mistake that occurs when the parents’ sperm or egg cells are formed. At one point in the formation, all the chromosomes including the two chromosome 6s pair up and swap segments. To pair up precisely, each chromosome ‘recognises’ matching or near-matching DNA sequences on its partner chromosome. However, throughout the chromosomes there are many DNA sequences that are so similar that it is thought that mispairing can occur. Although no-one has ever seen this happen, it is believed that when the next step - the exchange of genetic material, known as ‘crossing over’ - follows, it is unequal, looping out and excising the length of the chromosome that is lost in a 6q deletion.

This is part of a natural process and as a parent there is nothing you can do to change or control it. Children from all parts of the world and from all types of background have 6q deletions. No environmental, dietary or lifestyle factors are known to cause them. So there is nothing that either parent did before or during pregnancy that caused the deletion to occur and equally nothing could have been done to prevent it.

One way that a deletion and a duplication could theoretically arise during the formation of egg or sperm cells. On the left are two matching chromosomes, each split to the centromere and ready to pair and exchange segments. The shaded bars show similar sequences of DNA in the chromosome that enable correct pairing. But just above the centromere - in the short arm in the diagram - mispairing has occurred. When the chromosomes separate (right), the mispairing has given rise to two normal and two abnormal chromosomes, one with a deletion and one with a duplication.
**Could this happen again?**

The possibility that a couple will have another pregnancy affected by a 6q deletion depends on their chromosomes. If both parents have normal chromosomes, the 6q deletion in the child has in all probability occurred as a chance event, in which case the child’s test results will be marked *de novo* or *dn*. It is then very unlikely to happen again.

If a test shows that either parent has a chromosome rearrangement involving 6q, the chances of further affected pregnancies with similar or occasionally different abnormalities are high. Once the family translocation has been characterised, it will be possible to diagnose an unbalanced chromosome arrangement prenatally. Other family members should also be tested in case they, too, are carriers of the balanced form of the rearrangement. A clinical geneticist or genetic counsellor will give individual guidance to the family.
Support and Information

Unique lists external message boards and websites in order to be helpful to families looking for information and support. This does not imply that we endorse their content or have any responsibility for it.

This updated information guide 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. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. The guide was compiled by Unique and reviewed by Professor Robert Hopkin, Division of Human Genetics, Cincinnati Children’s Hospital Medical Center and by Unique’s chief medical advisor, Professor Maj Hultén, Professor of Medical Genetics, University of Warwick, 2007. Version 2. (PM)

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Facebook page for chromosome 6: www.facebook.com/groups/chromosome6

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