

Support and Information



Understanding Chromosome & Gene Disorders

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Chromosome 6 research project

The C6 project works with families to collect detailed information with the aim of linking specific disease characteristics with specific regions of chromosome 6.

<https://www.chromosome6.org/>

Facebook page for chromosome 6

www.facebook.com/groups/chromosome6

This 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, Cincinnati, Ohio, USA, by Dr George Tanteles MD, MRCPH, Specialist Registrar in Clinical Genetics, Nottingham University Hospitals Trust and by Professor Maj Hulten BSc, PhD, MD, FRCPath, Professor of Medical Genetics, University of Warwick, UK 2007. Revised 05/09. (PM)

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Understanding Chromosome & Gene Disorders

6q deletions from 6q25



Sources & references

The information in this leaflet is drawn partly from medical publications. The first-named author and publication date are given to allow you to look for articles on the internet in PubMed.

The leaflet also draws on *Unique's* database. When this leaflet was written, *Unique* had 67 members with a 6q deletion, of whom 46 had a pure 6q deletion with no other chromosome involved.

ARID1B syndrome

Changes (mutations) or loss of the *ARID1B* gene in the 6q25.3 band cause ARID1B syndrome. *Unique* publishes a separate short guide to this single gene disorder syndrome.

A chromosome 6q deletion means that part of one of the body's chromosomes has been lost or deleted. If the missing part contains important instructions for the body, some learning difficulties or disability, developmental delay and health problems may occur. How serious these problems are depends on how much of the chromosome has been deleted and where the deletion is.

Genes and chromosomes

Our bodies are made up of billions of cells. Most cells contain a complete set of genes. We have thousands of genes which act like a set of instructions, controlling our growth, development and how our bodies work.

Genes are carried on microscopically small, thread-like structures called chromosomes. We usually have 46 chromosomes, 23 inherited from our mother and 23 inherited from our father, so we have two sets of 23 chromosomes in 'pairs'. Chromosomes and genes are made up of a chemical substance called DNA.

Apart from two sex chromosomes (two Xs for a girl and an X and a Y for a boy), chromosomes are numbered 1 to 22, generally from largest to smallest. Each chromosome has a short (**p**) arm (at the top in the diagram) and a long (**q**) arm (at the bottom). In a 6q deletion, material has been lost from the long arm of one chromosome 6.

The chromosome may have broken in two places and the part between them is missing (an **interstitial** deletion) or it may have broken in one place and the part of the chromosome from the breakpoint to the end of the arm is missing (a **terminal** deletion).

Looking at 6q25

You can't see chromosomes with the naked eye, but if you stain them and magnify their image under a microscope, you can see that each one has a distinctive pattern of light and dark bands. Staining

6q25.2q25.3 deletion

Having studied four very young children with a deletion involving band 6q25, researchers from the Baylor College of Medicine, Houston, USA, have suggested that a 6q25.2q25.3 microdeletion syndrome exists. People with this syndrome have an unusually small head, developmental delay, unusual facial features and a hearing impairment. Some have absence of the corpus callosum, the band of nerve fibres that links the two hemispheres on each side of the brain (Nagamani 2009).

Information about four *Unique* members with a piece missing from 6q25 suggests that it may be too soon to define a syndrome. None of the *Unique* members has evidence of a permanent hearing loss and only one has incomplete formation of the corpus callosum.

Growing up

Information on puberty is available from *Unique* on one boy, who progressed normally from the age of 18. He left school at 18 and went on to college to study animal welfare and computer studies. He lived in a residential setting, not with his parents. Socially, he had his own friends as well as friendly relationships with neighbours and adult friends of his parents.

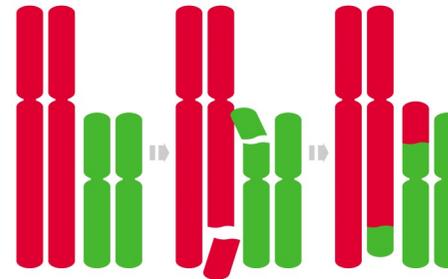
Why did the deletion occur?

Most 6q deletions occur when both parents have normal chromosomes. The term that geneticists use for this is **de novo (dn)**. A blood test to check the parents' chromosomes will show if this is the case.

De novo 6q deletions are caused by a change that has usually occurred when the parents' sperm or egg cells were formed. We know that chromosomes must break and rejoin when egg and sperm cells are formed but this only occasionally leads to problems.

The breaking and rejoining is part of a natural process and as a parent you cannot 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. There is nothing that either parent did before or during pregnancy that can be shown to have caused the deletion to occur and equally nothing could have been done to prevent it.

Some 6q25 deletions are the result of a rearrangement in one parent's chromosomes. This is usually a **balanced translocation** in which material has changed places between chromosomes but no material has been lost or gained and the parent usually has no difficulties with health or development.



Balanced translocation in one parent: material from the long arm of one chromosome 6 has swapped places with material from the short arm of another chromosome.

Can it happen again?

The possibility of having another pregnancy with a 6q deletion depends on the parents' chromosomes. If both parents have normal chromosomes, the 6q deletion is very unlikely to happen again. If a blood test shows that either parent has a chromosome change involving 6q, the possibility is increased of having other pregnancies with chromosome changes. Once a family chromosome change is known, a test in any future pregnancy can find out whether the baby's chromosomes are affected. A genetic specialist can give you more guidance.



Using their hands: fine motor and coordination skills

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 consistent mild to moderate delay in hand use and fine motor skills as there is in toilet training. This delay means that early intervention by occupational therapy to stimulate hand use is vital. An adult with a 6q25 deletion can wash himself, brush his teeth and dress himself with help putting on socks and shoes.

Speech and communication

Some delay in the emergence of speech and language is to be expected, but the extent is variable and probably reflects cognitive ability, although in one child speech and language were specifically delayed. A boy with a deletion between 6q24.3 and q25.2 and no learning difficulties was speaking in single words by 18 months (Tanteles 2007). While one five year old was communicating with vocal sounds and understood some familiar questions, another had good expressive language although he still had difficulty pronouncing some sounds of speech. One 17-year-old has very few words, while another adult has fluent, correct speech with a wide vocabulary, chatters constantly, can express himself and understand most things but can be repetitive. All children benefited from consistent high quality speech therapy.

Learning

Some learning support is likely to be needed but it is unlikely to be possible to predict the learning level from the karyotype. It seems that most youngsters have a mild to moderate degree of learning difficulty but at 9, a boy with a deletion between 6q24.3 and q25.2 has no learning difficulties (Tanteles 2007; Striano 2006). Individual children learn best through music and computer aids. They may learn better visually than aurally because they can set their own learning pace with visual material. One thirteen-year-old could find favourite websites on the computer; another youngster who was writing at the age of six, reading at seven and using a keyboard at 10, had a selectively excellent memory and was particularly good at number work. There is limited evidence that some children have musical talent and can sing with perfect pitch and rhythm, even if they cannot master the words of a song.

Behaviour

Most children are described as happy, affectionate, cuddly and sociable. They may take the initiative to come for a hug, love things going on and get bored with an unstimulating environment. Depending on the stimulus, they may be able to play for long periods with good concentration and may have good imaginative play or enjoy repetitive activities. Frustration can lead to self-injurious behaviour (hitting themselves, eye-poking, head banging) but they usually respond well to praise or direction. Among older people, there is some evidence of stubborn behaviour, periods of overactivity and restlessness and temper tantrums. In general, children may not cope well with stress.

- “ Enjoys laughing especially with daddy; waving toys especially with right hand - 10 months
- “ She loves to be with her family and other children, can play with toys for hours and cannot resist challenges like climbing onto chairs. Hugs and follows older children.
Likes to play with toys that can be dragged around. Sings 20-30 songs with perfect pitch and rhythm. Loves stimulation, something happening. Social, likes praise - 5 years
- “ Enjoys action figures; pretending; animals; dolls and teddies; children's TV; does not like motors. Can be disruptive due to over excitement - 5 years
- “ Loves to stand over a motorway and wave to cars; enjoys playing football, watching train DVDs, walking, cutting up raffle tickets. Can be cooperative - or stubborn - 21 years

reveals three bands within the 6q25 region, known as 6q25.1, 6q25.2 and 6q25.3. This leaflet tells you about deletions within these bands or terminal deletions with one breakpoint within these bands.

The missing piece of chromosome can be tiny or much larger. If it is large enough to be visible when magnified upto 1000 times under a microscope, it is called a **deletion**. Conventional chromosome analysis uses magnification to detect changes in chromosomes. Sometimes the missing piece is so tiny that it can only be identified using newer technology with tests such as FISH or array-CGH. It is then called a **microdeletion**. Smaller deletions generally remove fewer genes. The newer technologies can usually show whether particular genes or parts of genes are present or not.

The karyotype

Your genetic specialist can tell you more about what chromosome material has been lost. You will almost certainly be given a **karyotype**, a shorthand code that shows the bands where the chromosome has broken and rejoined. A band can contain many genes and depending on the technology used to find your child's chromosome deletion, the karyotype sometimes shows whether particular genes are present or not. But you will usually need to ask your genetic specialist for a full explanation.

Your child's karyotype may look very like another person's, from *Unique* or in the medical literature, or it may look exactly the same. But even in people with the same karyotype, the chromosome may have broken at a different point within the same band. This is one important reason why people with apparently similar karyotypes do not all have the same problems or features. Individual differences can be quite marked and it is very important not to make direct comparisons between your child and others. After all, each of us is unique.

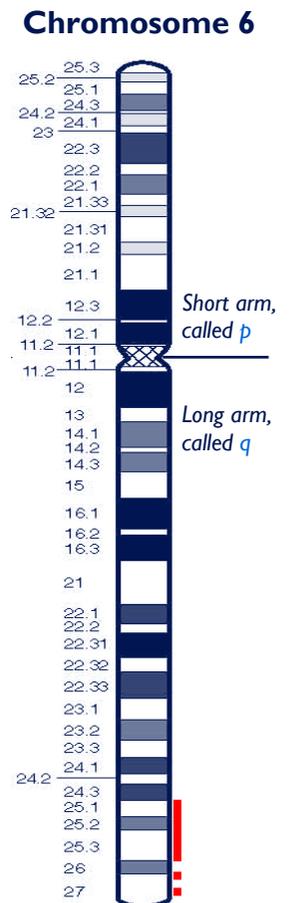
But some features and health problems are similar in people with a 6q25 deletion. This leaflet describes the things that are similar.

Around 40 cases with a pure deletion of the area marked in red on the diagram (right) are described, at least 26 from the medical literature and 13 members of *Unique*..

Most people have a single breakpoint and the end of the chromosome is missing, but a few have a small interstitial deletion within the 6q25 bands (see page 2).

The oldest member of *Unique* was 21 ywhen this information was compiled and the oldest person described in the medical literature was a woman of 37.

(Tanteles 2007; Bisgaard 2006; Striano 2006; Poyhonen 2005; Lukusa 2001; Sukumar 1999; Koh 1998; Pirola 1998; Hopkin 1997; Evers 1996; Rubtsov 1996; Treacy 1996; Meng 1992; Valtat 1992; Narahara 1991; Krassikoff 1990; Oliveira-Duarte 1990; Stevens 1988; Rivas 1986; Bartoshesky 1978; Liberfarb 1978; Milosevic 1975; *Unique*)



Most **pregnancies** were considered normal by the mother and, where this is known, by the caregiver. Some mothers commented on the lack of fetal activity, while one baby was active as a fetus and continued to be restless and waving his limbs as a newborn. In one pregnancy the level of amniotic fluid was low and there was some bleeding in the second trimester. Two babies were delivered by Caesarean section after experiencing fetal distress or a drop in heart rate during contractions. Ultrasound scan revealed enlarged ventricles (fluid-filled spaces) within the brain in one baby.

At birth

Range of birth weights at or near term: 5lb 11oz/2.575 kg to 8lb 6oz/3.8kg

While some babies had a lower-than-average birth weight, none was small for dates and more than one in five was above average weight. Apgar scores, rating babies' condition at birth on a scale of 0-10, ranged between 4 and 10 and this, together with feeding difficulties and babies' appearance generally alerted medical staff to a possible underlying problem. One baby was blue and spent the first day in an incubator, another was irritable, a third was restless and moved asymmetrically. One baby had a distinctive cry. However, some babies gave no cause at all for concern at birth and a problem was only suspected when development was delayed or growth in head size tailed off.

Feeding

Many babies with a chromosome disorder have initial feeding difficulties and the evidence from *Unique* and to a lesser extent from the medical literature is that babies with a 6q25 deletion are very likely to be affected. A significant number of babies in the *Unique* series developed feeding problems after the newborn period. One baby stopped feeding at three months; another gradually lost interest in food in the second year and severe eating problems developed by five years. In this case, eating problems correlated with her epilepsy and increasing medication appeared to help. One baby with a deletion between 6q24.3 and q25.2 and otherwise normal development was fed to the age of three by naso-gastric tube (a feeding tube threaded through the nose and down into the stomach); by five years he was feeding normally (Tanteles 2007).

Babies are likely to have difficulty establishing feeding. They may show no interest, be unable to suck effectively or to coordinate the actions of sucking with swallowing; they may feed but not so as to meet their nutritional needs. In some babies the problems are mild and temporary and it is possible to breastfeed. Other babies need long-term support and may need to be fed, as above, either through a naso-gastric tube or a gastrostomy tube (PEG, button) direct into the stomach.

Once children move on to solids, they may have difficulty with lumpy foods and need their feeds pureed for very much longer than other children. They may also have difficulty moving food from the front to the back of the mouth and need to have a spoon of food alternated with a drink. In some children the tongue position may make it difficult to spoon feed.

Gastro oesophageal reflux (GORD, GERD), where the stomach contents return up the food passage, may occur. Reflux raises a baby's risk of inhaling food 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

Outlook

The outlook for any child is determined largely by their clinical problems. Three babies in this group, all with serious medical concerns, are known to have died. Two died in the newborn period and a third child whose heart condition was not surgically corrected died at the age of two.

■ Hearing

The great majority of babies and children appear to hear without problems and there are no confirmed reports of children with a permanent hearing loss. As with all children, when young they will be prone to the temporary and fluctuating hearing loss caused by a build-up of fluid in the middle ear but there is no evidence that children in this group are more prone to this than other typically developing children.

■ Eyesight



Your child is likely to be offered a detailed ophthalmological examination. Many babies have been found to have abnormalities of the retina or macula at the back of the eye and at least four children had a significant vision defect, requiring vision therapy. In one child a defect of the macula was identified akin to macular degeneration, which affects central vision but typically leaves peripheral vision unaffected. One baby was described with a condition known as ocular albinism, in which the eyes lack melanin pigment.

Strabismus (squint) was also fairly common, affecting around a quarter of the children. This may 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 monitoring, treatment or surgical correction in others. Nystagmus, making the eyes move to and fro or up and down, was seen in one child, and may affect them particularly when tired. It should be carefully assessed but may be permanent.

Development

Sitting, moving: gross motor skills

The great majority of children with a deletion in this area can expect to develop more slowly than other children, although delayed development is not universal: one *Unique* member with a deletion between 6q25.1 and 6q25.3, 13 years old when this information was compiled, had entirely normal mobility and a boy with a deletion between 6q24.3 and q25.2 walked at 18 months and had normal mobility but some balance problems at age 5. In general, babies learned to roll over between five and 13 months, were able to sit up by nine months to three years, were walking in their third year and walking unsupported soon afterwards and climbing stairs from around their third year. Some babies never crawled but bottom-shuffled instead; those babies who did crawl achieved this between 12 and 18 months. One child had asymmetric development, with an obvious preference for his right hand side. Once moving, some children were highly active and mobile, although one child had a gradual increase in muscle tone throughout his first year. He was managed with daily stretches but by 19 months muscles in the upper and lower leg were slightly shortened and at the age of five he was walking with his knees slightly bent.

Children benefited from early physiotherapy. Many needed special seating in the early days.

■ Seizures

Seizures occurred in at least one third of children, but in some cases they were a one-off event or only occurred with a high temperature, and epilepsy was not diagnosed. First seizures occurred at a variety of ages between two months and eight years and there was no evidence from *Unique* that they were hard to control. In one child with diagnosed epilepsy, non-seizure activity was held responsible for behaviour and eating difficulties.

It has been suggested that there is a typical pattern of epilepsy associated with a 6q terminal deletions with signs such as vomiting and turning blue. However, overall, seizures are well controlled with or without medication (Striano 2006).

■ Heart

Defects in the structure of the heart are seen quite commonly in babies with a chromosome disorder and were found in around one third of babies. Some identified anomalies were simple and include a hole between the two upper (holding) or lower (pumping) chambers of the heart (atrial septal defect/ASD or ventricular septal defect/ VSD), persistence of a channel linking two major blood vessels leaving the heart that normally closes after birth (persistent ductus arteriosus/PDA), persistence of the foramen ovale, a connection between the two upper chambers of the heart that is open in the unborn baby but normally closes at birth and pulmonary stenosis, where the entrance to the artery that takes blood to the lungs is unusually narrow.

Other babies were born with a complex heart condition. One baby was born with an atrioventricular canal (AV canal/AVC), where there is a large hole in the middle of the heart connecting the two upper chambers (atria), the two lower chambers (ventricles) and a single atrioventricular valve instead of two separate valves on either side of the heart. In this defect, the valve often does not work properly and the large hole allows too much blood to flow to the lungs. Two babies were born with the complex anomaly known as Tetralogy of Fallot, involving both a VSD and an obstruction just below the pulmonary valve that decreases the normal flow of blood to the lungs, with the aorta shifted to the right so that it sits astride the VSD. Another baby was born with a small, third collecting chamber in the heart (atrial heart) into which the veins drain blood from the lungs. Having an extra chamber slows the passage of blood through the heart and may eventually lead to features of obstruction and heart failure.

Heart conditions will need monitoring, treatment with medication or surgical correction.

■ Kidneys

Four babies had an enlargement (hydronephrosis) of one kidney or both. There are many causes, including a blockage in the drainage of urine, reflux of urine from the bladder, a double ureter leading from the kidney to the bladder and a non-functioning, cystic kidney. Treatment of hydronephrosis depends on the cause.

■ Other concerns

One child had an **inguinal hernia**, a protrusion of part of the bowel through the inguinal canal (the channels through which the testes descend into the scrotum). A young girl had hair growth in the pubic area, suggesting possible premature puberty or adrenarche, where some characteristics such as body hair appear early but other signs of puberty (such as a growth spurt) do not follow.

One baby was born with an **imperforate anus** (absence of the normal anal opening). This is repaired surgically.

reflux problems are persistent, a gastrostomy tube can be inserted to allow direct feeding into the stomach until the baby is sufficiently mature to tolerate feeding by mouth.

Growing

Babies and children with a 6q25 deletion can be tall, medium or short for their age but the evidence suggests that while it is as common to be of medium height or short, only a few children are tall. Their height is usually proportionate but their head may be small relative to the rest of the body. High activity levels and a small appetite mean that most youngsters appear to be thin and wiry; one adult weighs four stone (25 kg). The rate of growth may not be steady; in one child, periods of normal growth were interspersed with periods when she did not grow at all; overall she was of normal height. There is some evidence that deletion of a gene known as *ESR1* at 6q25 may cause abnormally short stature (Bisgaard 2006).

Appearance

Doctors may notice in a baby what are known as 'dysmorphic features' which may or may not be obvious to a parent who may, however, notice that their baby does not resemble other family members. You may see similarities between your child and the pictures on the front of this leaflet.

Some of the most common features are seen in many babies and children with a chromosome disorder, while others are more specific to a 6q25 deletion. The most common features seen in young babies include a small chin and lower jaw that may recede from the upper jaw, low set ears that may be large, protrude or tilt backwards, a short neck, sometimes with loose skin, wide set, sometimes short or narrow eyes that may slant up or down and may have a tiny skinfold across the inner corner, a broad and usually flat bridge to the nose which may be beaked or bulbous, in particular the tip; high, rounded cheeks with on occasion a tiny hole just in front of the ears and sometimes thin, sparse hair. The face or part of it may look asymmetrical as may the head, which may be an unusual shape.

As for adults, the young man on the cover of this leaflet is 21 years old and there is a report in the medical literature with a photo of a 37-year-old woman with a deletion from 6q25.3 with a long face, wide mouth and small chin (Striano 2006).

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. In terms of hands, babies in this group have been described with short fifth fingers, long and sometimes tapering or very flexible fingers, webbed fingers, low placed thumbs, small hands, a single crease across the palm, either short or long final joints on the fingers and dry loose skin over the fingers. A child with a deletion between 6q24.3 and q25.2 had loose skin on the backs of his hands and feet and deep sole and palm creases. One child with a terminal 6q25 deletion was born with a serious hand defect involving a missing third finger on the right hand and a fusion of the second and third fingers on the left hand. Her feet were also affected, with the second and third toes on the right foot missing while the same toes on the left foot were fused.

Among other children, anomalies were mostly positional or could be managed with adapted footwear or insoles. The most common were rocker bottom feet (the sole is curved like a rocker), flat feet, overlapping or clenched toes, abnormally shortened toes, webbing between toes or feet that were curved inwards or outwards, as well club foot.

Concerns

	Numbers reported
■ Feeding difficulties	Most
■ Hydrocephalus	13/39
■ Seizures	At least 13/39
■ Heart conditions	Around 14/39
■ Sacral dimple	10/39
■ Minor genital anomalies	7/19 boys; 2/20 girls
■ Strabismus (squint)	Around 10/39
■ Spinal defect	7/39
■ Cleft palate	5/39
■ Hydronephrosis	4/39
■ Vision defect	4/39
■ Umbilical hernia	2/39
■ Diaphragmatic hernia	2/39
■ Choanal atresia	2/39

These conditions are explained in the text

Conditions you might see at birth

A **sacral dimple** (dimple or hole in the skin just above the crease between the buttocks) or a similar defect near the base of the spine was seen in 10/39 babies. The dimple 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 all-important. A sacral pit may be deep and even connect to the spinal canal. If there is any concern about this, your baby's spine will be imaged, usually with ultrasound or an MRI scan.

Occasionally, the end of the spinal cord that is usually free within the spinal column becomes attached to one of the surrounding structures. This is called a **tethered cord**, seen in one *Unique* child. 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. If necessary the cord can be surgically released to hang freely. At least three babies were also born with a prominent coccyx, where the base of the spine bulges slightly outwards. In itself this is most likely to cause a little discomfort while sitting. Two babies were born with spina bifida.

Minor anomalies of the **genitals** and the bottom area are often seen in babies with a chromosome disorder, especially boys. Seven out of 19 boys either had undescended testicles, very small genitals or both. 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. Two out of 20 girls had minor genital anomalies; in one, the hole for the bottom (anus) was unusually far forward, so that hygiene when changing nappies/ diapers is especially important; in another the vagina lips were small.

Five babies were born with a **cleft palate** (a split in the roof of the mouth), which usually needs surgical repair for feeding and speech development; none of the *Unique* series had a cleft palate, although many had an unusually high palate.

Two babies were born with an **umbilical hernia**, seen in other babies with a 6q deletion, but none in the *Unique* series was affected. This shows as a soft, skin-covered bulge at the umbilicus (navel, belly button) that can get 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, while a large hernia may need surgical repair.

Two babies were born with a **diaphragmatic hernia**, where the muscular wall separating the heart and lungs from the contents of the abdomen develops with a hole. Part of the bowel, stomach or liver take up space in the chest, potentially depriving the lungs and heart of room to develop properly. Immediate surgery is needed and the baby will need respiratory support until recovery from surgery. However, the consequences of a diaphragmatic hernia are not always recoverable and your baby's doctors may wish to discuss with you the wisdom of surgical intervention.

Two babies were born with **choanal atresia**, where the nasal passages are blocked by bone or tissue. One or both nasal passages can be affected. When only one side is blocked, a baby may not show many symptoms and so the condition may not be diagnosed for some time. When both sides are affected, a baby will have some difficulty breathing and so the condition is likely to be diagnosed and surgical repair needed soon after birth.

One baby was born with a **cystic hygroma**, a sac-like structure filled with lymph, most commonly in the head and neck area, often appearing as a soft bulge under the skin. It is usually removed by surgery.

Medical concerns

■ Head and brain

Most typically, head size among babies and children with a 6q25 deletion appears to be small (microcephaly). This judgement may be absolute or relative to the rest of the body. Some children also have an unusually shaped head and in a small number of babies there is evidence that some of the bony plates of the skull may fuse early; surgery to re-open the skull plates was undertaken in at least one child and considered in another.

Despite the small size of the head, hydrocephalus – excess fluid within the fluid-filled areas of the brain or enlarged fluid-filled areas – was seen in at least 13 babies or children. The cause of the fluid build-up was not always noted, but in at least two children it was caused by a blockage (aqueductal stenosis). A baby or child with hydrocephalus will be carefully evaluated and monitored. If treatment becomes necessary, excess fluid can be drained either through a permanent shunt that drains fluid from the ventricle to the abdomen or, in certain cases, by making a hole to allow drainage from a blocked area to an area with normal drainage.

A typical brain anomaly in those with a 6q terminal deletion is colpocephaly (in which the occipital horns - the rear portion of the lateral ventricles of the brain - are larger than normal), sometimes associated with an anomaly of the corpus callosum, the band of nerve fibres that links the two hemispheres of the brain (Striano 2006).

Imaging of the brain revealed further anomalies in some children, including the absence, thinning or delayed maturation of the corpus callosum and a delay in the myelination (insulation) process of the nerve fibres. What these findings may mean to an individual child is not always clear, but your child's neurologist will be best placed to interpret them.