21q deletions
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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. Like most other chromosome disorders, this increases the risk of birth defects, developmental delay and learning difficulties. However, the likelihood of problems developing depends very much on which genetic material is missing.

Chromosomes are the microscopically small structures in the nucleus of the body’s cells that carry genetic information. They can be stained so that each has a distinctive pattern of light and dark bands when viewed at about 1000 times life size under a light microscope.

Chromosomes come in different sizes, each with a short (p) and a long (q) arm. Apart from the sex chromosomes (two Xs for a girl and an X and a Y for a boy), they are numbered from largest to smallest approximately according to size from number 1 to number 22. Actually, though, chromosome 21 is the smallest chromosome, containing no more than 200 to 400 genes. The short (p) arm of chromosome 21 is very small and similar genes are available on other chromosomes, so it can usually 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.

**Chromosome 21**

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For common breakpoints, see page 15.
Main features

Deletion from the tip of the short arm to band 21q21
Many people with material lost between the tip of the short (p) arm up to and including the very first part of band 21q21 are healthy, have no learning difficulties or need only a small amount of support with their learning and go on to lead independent adult lives.

Some people with more material lost from 21q21 do, however, 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.

Deletion including band 21q22
Learning difficulties are fairly common and although the range is variable, they are generally at least moderate, so learning support is needed.

- At birth, the baby’s head may be very small (microcephaly) and it may grow slowly. This does not need treatment.
- Joints may be stiff and held in an unusual position. Physiotherapy, splinting and surgery help to make them more flexible.
- The muscles of the body may feel either too tight (hypertonia) or too floppy (hypotonia). Physiotherapy helps.
- 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.
- There is a wide range of other possible birth defects. The paediatrician will check your child carefully for these.


First signs

Unique’s experience is that the first signs of a chromosome disorder that led to the diagnosis were varied and children were diagnosed at very different ages. Two babies (with a q11.2q22.1 and a 21q22.1q22.3 deletion) were identified at birth, in one because of overlapping fingers and toes, poor weight gain and sucking ability as a newborn. One baby with microcephaly identified during pregnancy was diagnosed after her development was very slow; a five year old with a deletion from 21q22 was diagnosed after he developed seizures. A teenager with a 21q21q22.1 deletion was diagnosed at 16 after she developed mental health problems in response to increased academic demands (Takhar 2002; U).
Pregnancy
Families’ memories of pregnancy are quite varied. In some, the pregnancy was uneventful and this was particularly true when the deletion did not include band 21q22. Among three families with involvement of band 21q22, one baby was born at 34 weeks, one at term and one at 42 weeks. One mother reported lack of fetal movement, another had a large amount of amniotic fluid and a third had a sense that her uterus did not grow enough to accommodate the baby.

The newborn period
Unique’s experience is that most babies with a loss including 21q22 experienced problems in the newborn period. Apgar scores (which rate a baby’s well-being at birth on a scale of 0-10) were typically between 4 and 8 one minute after delivery and 8 to 9 at five minutes. Weak sucking was typical and some babies had difficulty establishing independent breathing. One baby had a pneumothorax, where air leaks into the chest cavity surrounding the lung. One baby with a deletion from 21q11 to 21q21, excluding the 21q22 region, had particular difficulties establishing independent breathing and was thought to have had minor strokes before birth.

Facial appearance
No single facial feature is usually a hallmark of any particular chromosome disorder, but a pattern of so-called ‘dysmorphic’ features is often typical. Doctors look for these characteristics because they can help them in their search for the correct diagnosis. They usually do not create a problem for the child but you may find that if your baby with a 21q deletion is not your first, you notice some facial features that are unlike the rest of the family. Your baby’s head may be unusually small and the back of the head may stick out noticeably (prominent occiput). The eyes, which may be small, can have an unusual upward or more commonly downward slant, the nose may be quite broad from the root between the eyes to the tip and the groove between the nose and the mouth (philtrum) unusually long. Some babies have a very wide, large mouth with downturned corners. Some have a small chin and lower jaw. A baby’s ears may well be rather large and placed low on the head, and sometimes rather simply formed. However, there is quite a lot of individual variation and other features noted by Unique families include a flat bridge to the nose and large tear ducts that easily become infected (Huret 1995; U).
Growth
Most medical texts say that babies with a 21q deletion are usually rather small at birth, with a weight and length that would put them in the lowest ten per cent of the population for size. The Unique experience is of quite varied growth patterns. While most children remain on the short side, they are not usually extremely short. Some children are exceptionally thin and have great difficulty in putting on weight. As an example, a nine-year-old girl with the height of a seven-year-old weighed 17 kilos (38 pounds), the average weight of a 3-year-old. Others are unusually tall: a boy with a deletion from 21q22 to the end of the chromosome was long at birth, remained tall and at the age of 17 was 196 cm tall (6’ 5’’). (Roland 1990; Huret 1995; U).

Food and eating
Unique’s experience is that babies and children who have deletions that do not include band 21q22 feed well from birth and only have mild difficulties, if any, with feeding. They may however find it difficult to put on weight. The feeding difficulties they encounter are similar to those in any group of babies without a chromosome disorder, such as mild gastro-oesophageal reflux (where the contents of the stomach return up the food pipe and may be vomited) and messy eating.

Among babies and children with a deletion including band 21q22, difficulties are more common and more severe, and families may benefit from feeding support, particularly as many babies do have difficulty in putting on weight.
Although progress through the normal feeding stages is usually delayed, children usually eventually eat well, choosing from a healthy range of foods. In newborn babies, sucking is often weak and not enough to meet nutritional needs. Some babies have difficulty co-ordinating the actions of breathing, sucking and swallowing and two out of four babies have been lactose intolerant. Feeding expressed breast milk from a bottle with an adapted teat may however be successful and your baby’s dietician or infant feeding counsellor will guide you. The move from a bottle to a spouted cup may come late and not be achieved until the school years. Transition to solids may also be late and many babies accept new textures, lumps and chewy foods slowly, needing their foods liquidised or mashed well into childhood. The resistance to chewing may remain throughout childhood and is partly caused by low muscle tone in the mouth. Speech and language therapists specialise in eating skills as well as speech and language. Occupational therapists can help with feeding skills and advise on adapted cutlery.

Constipation
Constipation is a common concern in young children with chromosome disorders. There are usually multiple causes, including a small appetite, low fluid intake, little physical activity and reluctance to bulk out foods with fibre. Family solutions (such as an ounce of prune juice in a bottle of milk, warm baths) may work, but if they do not, your GP can prescribe both stool softeners and stimulants. The experience of Unique is that children with 21q deletions tend to outgrow their constipation by school age.
Food and eating - families say:

“Either he swallowed too slowly, or my milk came out too fast, but I always remember feeding times as less than satisfactory. At the beginning we were often admitted to hospital as John’s lips would go blue, or I would have to turn him upside down as he seemed to be choking. It would always seem to be such a long winded, drawn out affair – 21q22 deletion.

“Feeding was very slow and mostly baby food but by four she would suck on a biscuit and a little toast. By 5, she was eating liquidised school dinners but still mostly baby food at home, and she had progressed from a bottle to a beaker. At the age of 8, her food was still liquidised but she would eat crisps, chocolate and bread & butter given to her. She would suffer a lot with trapped wind and could get quite angry and violent until she had released it - 21q22.1-q22.3 deletion.

“She remained on baby food past her first birthday because she could not tolerate textured food. She gradually went on to soft foods such as yoghurt, apple sauce and pasta. At about 5 years of age she started eating textured foods and now eats almost everything. She still does not like chewy food – 21q11.2q22.1 deletion, age 8.

Learning and schooling

For people with a deletion that does not include 21q22, it is hard to predict in babyhood the outlook as far as learning is concerned. There are many instances in the medical literature and within Unique of people with a deletion that ends within band q21 or at q22.1 who have not apparently had any learning difficulties and have had a professional career. Others have had slight learning difficulties, particularly in coping with the demands of secondary education. A smaller number have had a moderate learning difficulty and needed support with their education. In one family with two children with a 21q11.2q21.3 deletion, one child had slight learning difficulties and the other had moderate difficulties.

For people with a deletion that includes bands 21q22.1 to q22.3, more difficulties with learning can be anticipated and children will usually benefit from special education (Chettouh 1995; Huret 1995; Chen 2004; U).

Learning and schooling - families say:

When the breakpoint is no further than 21q22.1

“Moderate learning difficulties and attends a mainstream school. At 14, she can read most books and newspapers. Her best subject at school is drama, her weak areas are physical education, English, drawing and maths. She learns best in a small group - 21q11q21 deletion.

“The gap with her peers has been small at primary school. She learned to read and write at the age of seven. At the age of 10, she had the numeracy skills of an 8 year old and
Communicating and speaking

Generally, children have a delay in speech and language skills that is in line with their level of learning difficulty. This means that 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. However, one child with a 21q11.2q22.1 deletion had delay in all areas of development except for speech, using words first at 9 months and at age 8, expressing herself well except when explaining how she feels.

Another baby with a 21q22 deletion had a tongue tie tethering the tongue to the base of the mouth. This can be cut in a straightforward surgical procedure.

In one family where two children had a 21q11.2q21.3 deletion both had a tendency to shout. The children had different levels of speech fluency, with one a good conversationalist at the age of 12, while the other had limited understanding, immature pronunciation and word choice and a flat voice.

Among children with a 21q22 deletion, speech typically takes longer to emerge.

“\textbf{At the age of 5, she has no speech yet, but she communicates in her own way. She can’t tell you in words what she wants but she will express displeasure or push away things she doesn’t want and will express joy for people or things she likes or wants.}
Sitting, walking, moving

Although it is certain that children with a 21q deletion are more likely to be late in sitting, walking and running than children without a chromosome disorder, some are quite mildly affected.

Underlying the delay in mobility is an alteration in normal muscle tone, so that the muscles either feel too tight and tense (hypertonia) or too floppy and loose (hypotonia). Most children with a 21q deletion have a mixture of hypertonia and hypotonia. Additionally, many children with a 21q deletion are born with one or more bent joints that do not have a full range of motion. Children with hypotonia are liable to have extremely mobile joints that are hard to control. Physiotherapy will improve both the muscle tone and the joints but may not achieve full correction and stiffness in the joints and an unusual walk may continue throughout childhood. Bent joints may also be treated with splinting and plaster casting to increasingly adjust to a more normal position, but the cast itself may then delay a child’s mobility, especially where more than one joint needs casting. Occasionally surgery is needed. In at least one Unique member, physiotherapy achieved a completely normal level of mobility by the age of seven (Roland 1990; U).

Babies learned to roll over between 3 and 15 months and to sit between 6 and 8 months. The age at which walking began varied widely among the two groups of children: where 21q22 was not involved, walking was only slightly delayed. Where it was partly or wholly lost, walking was markedly delayed and children only achieved mobility at school age. Some children were not walking and used a wheelchair. Among those who were walking, gait might remain unsteady so the child wore a protective helmet in case of falls. Some children needed support (splints or orthoses, or a walking frame) in the early stages of walking and this was especially true where 21q22 was deleted. Some individuals walked at first on tiptoe due to tightening of the tendons between the heel and the knee or the knee and the hip. This can be treated with support boots or splints or by casting to gradually lengthen the tendon or if this does not achieve enough correction, by releasing the tendon surgically (Korenberg 1991; U).

An additional concern for some children who have had low muscle tone in the upper body is an increasing spinal curve (scoliosis). Among Unique families, no child was seriously affected or needed more than regular monitoring.

Using their hands

Most children can expect some delay in using their hands and in hand eye co-ordination. In children with an intact 21q22, the delay may be slight and show only as clumsiness or specific difficulties with hand eye co-ordination that become important once the child is at school. Among children with a deletion of 21q22, there appears to be a tendency to keep the hands clenched in a fist. Home treatment - rubbing the palms and gently, regularly opening the hands - and occupational therapy should be available from the earliest days (U).
Medical concerns

Respiratory infections
Respiratory infections are the most common health concern among Unique families and were the most frequent reason for babies and children with a 21q deletion needing unplanned hospital treatment. Respiratory infections occur more commonly among children with a chromosome disorder than among other children and there is no reason to believe that they are caused specifically by the 21q deletion. Families reported that in some babies, infections of the lower airways (chest infections, pneumonia) were common from birth while in others they were most prevalent during the pre-school years. Children caught colds easily and often and slight infections turned readily and rapidly into a more serious chest infection. This tendency improved but did not disappear with age, being able to sit upright and better mobility. One child remained wheezy and, coupled with house dust mite allergy, developed asthma (Matsumoto 1997; Tinkel-Vernon 2003; Marafie 2005; U).

Seizures
Each of the three Unique children with a simple deletion that included band 21q22 has experienced seizures, although no epilepsy was seen in others. In one child the seizures followed two accidents at the age of 6 and were apparently outgrown within two years; another child developed them from the age of one and is still affected at the age of five with incomplete medication control; a third child with a history of febrile convulsions as a baby developed seizures at puberty when 10 years old and as a teenager is severely affected. Each of the Unique children with seizures also had learning difficulties, but one child described in the medical literature with seizures had normal intelligence (Korenberg 1991; U).

Joints
There is a marked tendency for babies with a 21q deletion to be born with one or more joints held in a bent position and unable to be fully flexed in all normal directions. The technical term for this is a contracture and when many joints are affected, the condition is known as arthrogryposis. It may be possible to identify this on an ultrasound scan before birth and it may progress after birth. Treatment can include physiotherapy (physical therapy) to gently stretch the joints, splinting and casting to progressively stretch the joints and to hold them in place and possibly surgery to release the fibrous strands constricting the joints. A tendency for the joints to dislocate easily has also been seen in one Unique member with a deletion including 21q22 (Huret 1995; Marafie 2005; U).

Blood disorders
Specific anaemias and blood disorders have been described in children with a deletion that involves 21q22. A mild degree of thrombocytopenia can occur, where
levels in the blood of the platelets that help to form blood clots are abnormally low. This means that cuts and nosebleeds take longer to clot and bruises can be extensive and in children with 21q deletions, this has been seen even when platelet levels are normal. Within Unique, of 11 children, one girl with a 21q22 deletion was severely anaemic at birth and has a low platelet count (Vogels 1994; Endo 1969; U). An inability to absorb vitamin B12 has also been described, but it is not certain that this was caused by the 21q deletion (Celep 1996).

**Heart conditions**
The medical literature cautions that structural heart defects are common in babies with a 21q deletion, affecting as many as three babies in four with the potential for serious problems such as 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), underdevelopment of certain heart valves (tricuspid atresia) or the complex condition known as Fallot’s tetralogy. However, Unique’s experience is more positive and none of its 12 members has a 21q deletion that has caused more than a heart murmur that on investigation has turned out to be harmless (Pai 2001; U).

**Brain**
A gene situated at 21q22.3 is known to be involved in the normal development of the forebrain in the embryo into two hemispheres (the left and right halves of the brain). Disruption of the gene’s natural expression causes holoprosencephaly. Holoprosencephaly can be extremely mild (showing, for example, as absent glands for smell) or more severe, affecting the brain’s function. It also usually affects the development of the face, but this may not be readily seen (Estabrooks 1990; Huret 1995; Muenke 1995; Pai 2003; Chen 2004).

Another feature of brain development that is sometimes seen in children with a 21q deletion is thinning or absence of the corpus callosum, the broad band of nerve fibres that links the two sides of the brain (Theodoropoulos 1995).

In general, however, the brain structure studies published so far are not detailed enough and although these anomalies have been reported repeatedly in the medical literature, the picture from Unique is more hopeful as none of the 12 children with a pure 21q deletion had a reported brain anomaly. However, this depends on the region deleted (Korenberg, personal communication; U).

**Kidneys**
The medical literature contains many reports of babies with missing kidneys or kidneys that have not developed correctly. However, Unique’s experience is less bleak: none of the 12 children with a pure 21q deletion had a structural defect of the kidney, although one girl had a mild enlargement of one kidney (Huret 1995; U).

**Anaesthesia**
The gene for superoxide dismutase, a scavenger of radicals, is presumed to be sited at 21q22.1. Individuals with only a single copy of this gene produce around half the superoxide dismutase of other people and during anaesthesia this raises their risk of
oxygen toxicity (Ackerman 1988).

Endocrine
A girl has been described with hypothyroidism at birth (low level of thyroid hormone, treated with thyroxine) and a 21q11.1q22.1 deletion (Ahlbom 1996).

Genitals
Unusual genital features have been repeatedly described in the medical literature, ranging from undescended testicles at birth in boys to ambiguous genitalia in babies of either sex. The only problem reported among the Unique series was undescended testicles in one boy, which were brought down and tethered in the scrotum in a straightforward surgery known as an orchidopexy (Huret 1995; Pai 2001; Chen 2004; U).

Ears and hearing
Children with chromosome disorders have a higher rate of diagnosed temporary hearing loss caused by glue ear than other children, partly because doctors are alert to their need for good hearing and are ready to act rapidly to correct any problems by having fluid drained from the middle ear and by inserting tubes (grommets) to equalise air pressure on either side of the ear drum. There is no evidence that hearing problems are particularly common and the only report in the medical literature of permanent hearing loss may not be caused directly by the 21q deletion (Wakui 2002; U).

Eyes and vision
Despite reports in the medical literature of structural eye defects affecting the front portion of the eye, of very small eyes and of cataracts, children within Unique have not had serious vision problems and the problems experienced have not formed any particular pattern. One child is reported as extremely short sighted, another has moderate short sight, one is very long sighted and has a squint (strabismus) while a fourth has visual processing difficulties that mean he lacks depth perception. One child is reported as showing an intense dislike of sunlight and as a teenager is being investigated for glaucoma, a rise in pressure within the eye (Houston 1981; Joosten 1996; U).

Teeth
Many children with a chromosome disorder have dental problems. The causes are multiple and include disrupted development before birth, low muscle tone in the mouth and tongue that affects the eventual shape of the mouth and dietary issues, including a longer than normal sucking phase, difficulty with tooth cleaning and relatively low fluid and low fibre intake.

Among children with a 21q deletion, disrupted facial development associated with holoprosencephaly may affect the development of teeth. Among Unique’s members with a 21q deletion, the failure of baby teeth to fall out to allow adult teeth to come through was common and teeth had to be removed under anaesthetic. Two children had large and protruding central top teeth and most children either had wide gaps between their teeth or very crooked teeth.
Therapies
Children will benefit from early assessment and access to the major developmental therapies – physiotherapy (physical), occupational therapy and speech and language therapy. Ongoing physiotherapy is vital to maintain and if possible increase flexibility, as well as building up muscle strength and developing co-ordination. Additional therapies, such as hippotherapy (riding for the disabled) are enjoyable and helpful in particular for children who do not achieve independent walking.

Experience with puberty
No specific pattern of puberty has emerged from Unique’s experience, but behaviour difficulties and mood disturbances have been more noticeable.

Behaviour
Children with speech and language difficulties frequently have problems with behaviour, due to frustration. Additionally, some children with 21q deletions have a very short attention span and some hyperactive behaviours. Behaviour management from early childhood helps to support parents in dealing with their child’s difficulties. Some children have shown extreme temper outbursts, but overall Unique has not found a distinct pattern to children’s behaviour.

Families say …
“**She has not changed much at all. She is very angry and aggressive and then over loving** – age 14.
“**Puberty has taken a long time already. We are still going through it** – age 17.

“**He can fly off the handle at no notice, and be very spiteful and bullying but when questioned never knows why he has been nasty. He has had an anger management course but has recently been given to throwing things in a rage** – age 12.

“**She is loud, can be quick tempered and is both happy and friendly and very shy. She will torment her brothers until they retaliate, then sit back and enjoy watching them get told off** – age 10.

“**She can become very aggressive, grabbing your face or anything nearby, getting upset and crying. We think this is caused by having a lot of trapped wind. On the other hand she comes over excited, running around and laughing** – age 9.

“**She will scratch, kick, hit and bite others as well as herself. When this happens we usually move her away from others or try to distract her with something if she’s doing it to herself** – age 5.

Mental health
There is some evidence that some people with a 21q deletion may need particular support to maintain their mental health. A 16-year-old with a 21q21q22.1 deletion and no other specific symptoms developed schizophrenia in response to increased academic pressures at her mainstream school but was successfully treated with medication. The researchers pointed out that she was missing a gene (known as APP) which raises the risk of early dementia in people who have an extra copy of it and have Down’s syndrome (Takhar 2002).
Among Unique’s membership, two children have needed mental health support. One girl developed acute separation anxiety and obsessive compulsive disorder in early childhood (U).

**Independence**

There are descriptions in the medical literature of people with a 21q deletion leading fruitful and self-sustaining adult lives and this is particularly true of people with a deletion that does not involve 21q22. Others with a 21q deletion will need some support throughout their lives. Although it is difficult to predict the level of support with any certainty in early childhood, the picture will become clearer as your child’s skills develop.

Unique’s experience is that among those with a 21q deletion that does not include band 21q22, toilet training is successful between 2 and 4 years and children master the tasks of daily living such as dressing and eating with usually no more than adult supervision (Chettouh 1995; U).

**What do children enjoy?**

- *Music and karate - age 14.*
- *Playing with a gameboy but without really knowing what he is doing; making things with dough, computer and cooking easy recipes - age 12.*
- *The lack of concentration is inhibitive but she will colour, watch TV or a video for short periods, she likes dancing to music and knows the tunes but the words never come out right - age 10.*
- *Playing with her little dog, playing in her doll house with her sisters, jumping on the trampoline and going for walks - age 8.*
- *She carries her fizz doll or a soft rabbit around. She loves music, watching the Tweenies, cooking programmes, going for walks, swimming and the family trampoline - age 9.*
- *She has a few people she loves to interact with and likes anything with lights and music - age 5.*
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 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.

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. Each of the 46 chromosomes first doubles lengthwise into two strands that are held together at the point where the short and long arms meet, known as the centromere. The chromosomes then arrange themselves in 23 pairs, with pairs lying alongside each other. The two members of each chromosome pair ‘recognise’ each other because the DNA sequence ladder that comprises them is in a similar order. However, when a small region of DNA on a chromosome has a twin region of DNA located further down the same chromosome, the pair of chromosomes may not align correctly. Usually, after chromosomes pair, the members of a pair exchange segments of DNA with their pair-mates, in a process known as crossing-over (recombination). After this point, the chromosome strands repel each other but are held together at the cross-over points known as chiasmata. Deletions can arise during this process when the chromosomes have lined up incorrectly. An unequal cross-over means that the exchanges are not equal between the members of a chromosome pair. In this case, a piece of one chromosome can loop out and be lost from the middle of the chromosome (interstitial deletion) or from the end of the chromosome that then ‘heals’ (terminal deletion).

Why did it 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 person in the family with rearranged chromosomes. The only way to know if the disorder is inherited or not inherited (when it is called de novo) is for the parents’ chromosomes to be checked and the results explained to you.

If the check reveals a structural rearrangement of one of the parents’ own 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 (Roland 1990; Chettouh 1995; Wakui 2002; U).

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. In families where one parent has the same 21q deletion as the child, the risk of it being passed on again can be as high as 50 per cent.
Some medical references

A full list including all the references in the List of deletions is available from Unique. Am J Med Genet A. 2003 Jul 1;120(1):142-3. PMID: 12794708 Patient with a deletion of chromosome 21q and minimal phenotype. Tinkel-Vernon et al


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 Dr Julie Korenberg, Cedars-Sinai Medical Center, Los Angeles, California, US, and by Professor Maj Hultén BSc PhD MD FRCPA, Professor of Reproductive Genetics, University of Warwick, UK 2005. [PM]

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