Understanding Chromosome & Gene Disorders

2q37 deletion syndrome

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What is the evidence?

Since the first report appeared in a medical journal in 1989, 74 babies, children and adults with a pure 2q37 deletion - involving no other chromosome - have been described in medical journals (Leroy 2012). Details of 66 people with a 2q37 deletion were published in 2004 by a US geneticist, Dr Kari Casas (Casas 2004). In 2007, Dr Casas and Dr Rena Falk from the University of California published a thorough review of the syndrome (Falk & Casas 2007).

In 2013, Unique had 88 members with a pure 2q37 deletion, ranging in age from newborn to 44 years. Following a general survey in 2003, and a survey of adolescents and adults in 2010, 23 families with a child under 14 completed a detailed survey of their child's health and development in 2012. This survey is referenced Unique. In April 2013, the 2q37 deletion Facebook group had 167 members.

The text contains references to articles in the medical literature. 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 (www.ncbi.nlm.nih.gov/pubmed). If you wish, you can obtain most articles from Unique.

2q37 deletion syndrome

2q37 deletion syndrome is a well defined chromosome condition. People with the syndrome have lost a small but variable amount of genetic material (DNA) from near the end of one of their two chromosome 2s. This affects their development, but how much they are affected, and the ways they are affected, can vary a lot. Researchers have repeatedly found striking differences between people who have lost almost exactly the same DNA.

When a particular set of features occurs as a result of a single cause in a recognisable and consistent pattern in enough people, the condition is called a syndrome. The main features of a 2q37 deletion occur in this way, so the disorder is known as 2q37 deletion syndrome.

How common are 2q37 deletions?

We don't know yet, but 2q37 deletions are found quite commonly among children who need support with their learning (Falk & Casas 2007).

Genes and chromosomes

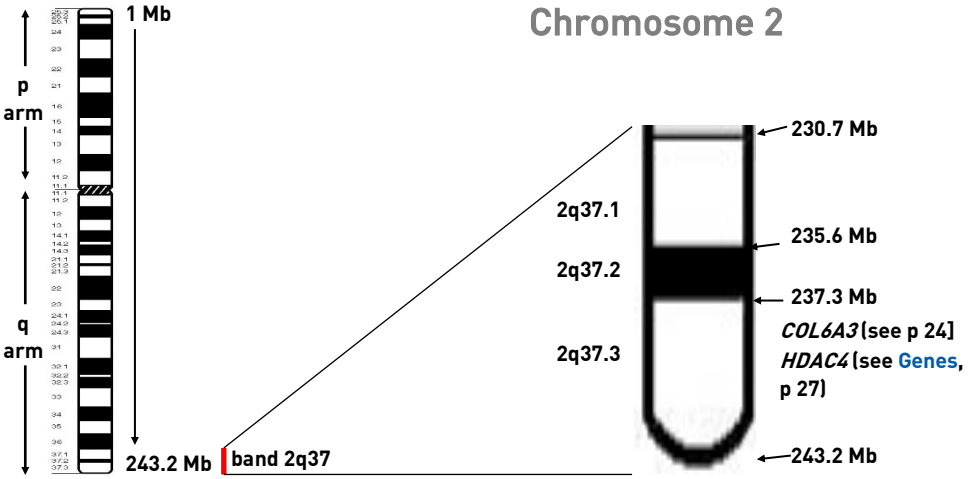
Our bodies are made up of trillions of cells. Most of the cells contain a set of around 20,000 different genes; this genetic information tells the body how to develop, grow and function. Genes are carried on structures called chromosomes.

Chromosomes usually come in pairs, one chromosome from each parent. Of the 46 chromosomes, two are a pair of sex chromosomes: (two Xs for a girl and an X and a Y for a boy). The remaining 44 chromosomes are grouped into 22 pairs and are numbered 1 to 22, approximately from largest to smallest. These are called autosomes. Each chromosome has a short (p) arm (from petit, the French for small) and a long (q) arm (*see* diagram).

Looking at chromosome 2q37

Chromosomes can't be seen with the naked eye, but if they are stained and magnified under a microscope, each one has a distinctive pattern of light and dark bands. In the diagram of chromosome 2 on page 3, you can see the chromosome bands are numbered outwards from the point where the long arm meets the short arm. 2q37 is at the bottom, divided into three bands – light (37.1), dark (37.2) and light (37.3).

Each band contains millions of base pairs of DNA. Base pairs are the chemicals in DNA that form the ends of the 'rungs' of its ladder-like structure. One band 2q37 has a little over 12 million base pairs. This sounds a lot, but is actually quite small: the DNA in 2q37 on one chromosome 2 is about 0.2 per cent of the total in each cell.



Genetic testing

Looking at chromosomes under a microscope, it may be possible to see the genetic material that is missing, if the piece is large enough.

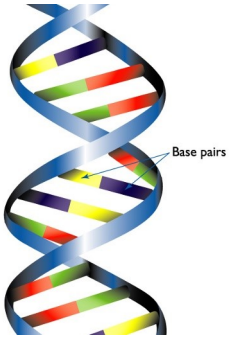
Molecular DNA technology gives a more precise understanding of the size and position of the deletion. This is important as scientists identify genes and pinpoint their location on chromosomes.

Techniques that are commonly used include FISH and microarrays:

- Fluorescence *in situ* hybridisation (FISH) uses fluorescent dyes to visualise under a microscope the number of copies of small sections of chromosomes. Unique publishes a separate guide to FISH

However, rare chromosome disorders may be caused by subtle changes in the chromosomes that are too small to see using a microscope.

- Microarray comparative genomic hybridisation (array CGH) is a sensitive technique which shows gains (and losses) of tiny amounts of DNA throughout the chromosomes. Array CGH identifies duplicated, disrupted or absent DNA. Unique publishes a separate guide to array CGH.



1 base pair = bp
1,000 base pairs = 1kb
1,000,000 base pairs = 1Mb

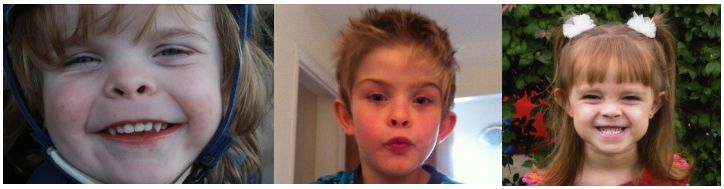
A person's chromosome make up is called his/ her karyotype. Someone with a 2q37 deletion might have a karyotype that looks like one of these three examples:

1. 46,XY,del(2)(q37.1) This result shows that the expected number of chromosomes [46] were found. It also shows that an X and a Y chromosome were found, so this is a boy or man. del(2) means there is a deletion from chromosome 2. (q37.1) shows the band in the chromosome where the break was found; in this case, the DNA is missing from this point to the end of the chromosome.

2. arr[hg19] 2q37.3(239836918 -243016613)x1 This result shows that a technology known as array comparative genomic hybridization (arr cgh) showed that only one copy (x1; the normal copy number is two) of part of the band known as 2q37.3 was found. hg19 tells you which version of the human genome was used to make these measurements. At present, hg19 is the latest version. The first base pair missing is 239836918 and the last is 243016613. By taking the first number from the second, you can work out that there are 3,179,695 missing base pairs, or about 3.18 Mb of missing material.
3. 46,XX,der(2)t(2;14)(q37.2;q32.33)mat About five per cent of people with a 2q37 deletion have a translocation where DNA has swapped places between two chromosomes. This can create a missing bit of one chromosome and an extra bit of another chromosome. This example shows someone with the expected number of chromosomes [46]. It also shows that two X chromosomes [XX] were found, so this is a girl or woman. der(2) means that there is an altered (derivative) chromosome 2. t(2;14) means that there is a translocation (t) between chromosomes 2 and 14 (2;14). (q37.2;q32.33) means that chromosome 2 has broken in the 2q37.2 band, and DNA is missing from that point; chromosome 14 has broken in the 14q32.33 band, and there is extra DNA from chromosome 14. mat means that the translocation has come from the mother. If it came from the father, it would say pat. Unique publishes a separate guide to **Balanced Translocations**.

Does everyone have the same size 2q37 deletion?

No, they don't. The chromosome can break anywhere in the bands called 2q37.1, 2q37.2 or 2q37.3 so people have different sized pieces of chromosome missing.



Left to right:
child with
2q37.1 deletion;
2q37.2 deletion;
2q37.3 deletion.

A normal variant in the general population

At the very tip of chromosome 2 there is material that can be lost without doing any harm at all. At least one person in 20 in the general population has lost a tiny bit of this material. As far as we know, this doesn't make them any more likely to have a child with a 2q37 deletion (Falk & Casas 2007; Leroy 2012).

Is there any difference between the effect on a girl and on a boy?

Not so far as anyone knows. The 2q37 deletion does seem to be more common in girl babies than boy babies, with 42 girls to 26 boys described in the medical literature. Within Unique there are 63 females to 25 males (Leroy 2012; Unique).

Are there people with a 2q37 deletion who have developed with no speech, behaviour, learning or health difficulties?

People with a 2q37 deletion will usually need help with their development and they may also have health and perhaps some behaviour difficulties. However, there are some young adults known to Unique and the Facebook group with only mild difficulties; at least one holds down a job. In 2013, a paper was under review describing some people with a 2q37 deletion who have normal cognition.

I wish ..

.. I wish someone had said to me that it's OK that my child has a genetic disorder. Instead I feel like everyone was trying either to get her to be 'normal' or convince me that I should just 'give in because she wasn't going to amount to anything' (actual words from a pediatrician we no longer see). 2q37.1q37.3 deletion, 19 months

.. I had known that doctors don't know everything. They said she would be short and she is only 1 inch shorter than me. Just because she has a chromosome abnormality does not mean she will not achieve what she wants to achieve. 2q37.1 deletion, 11 years

.. I could make every professional encountered read up on this deletion first to gain some understanding of what we are dealing with. 2q37.3 deletion, 6 years

.. I'd known that knowledge does not mean wisdom. 2q37 deletion, 26 months

.. her deafness had been diagnosed a good deal earlier, not at 2½ years old. 2q37.2 deletion, 3¾ years

.. I'd had more information and known where to get it, including Unique. He was diagnosed at birth and we knew barely anything except the name of the chromosome and that he was missing it. 2q37.1 deletion, 4 years

Main features of 2q37 deletion syndrome

In young babies

- Low muscle tone, causing floppiness (hypotonia)
- Feeding difficulties
- In some, health concerns

Later on

- Developmental delay - therapies help
- Low muscle tone and loose joints - physiotherapy is helpful
- Characteristic changes in facial appearance, hands and feet
- Learning disabilities - support is usually needed
- Tendency to gradually put on too much weight - exercise and a good diet help
- In some, difficult behaviour and some features of an autism-like disorder - but many children are very sociable
- In a few, seizures - these usually respond well to treatment
- Eczema, asthma, and frequent chest and ear infections are common

These features are now discussed in more detail.

First signs

There is great variability

First signs vary a lot. Many babies are born healthy but have 'soft signs', such as initial difficulty feeding and gaining weight or unusual floppiness, or some unusual facial or hand features. Later on, delayed development is a common first sign, usually together with other indications such as bad eczema or persistent feeding difficulties. When development is delayed, parents often have a sense that something is wrong but can't pinpoint the problem. A minority of babies are unwell from birth with, for example, breathing difficulties or a heart or gastrointestinal condition, or they have unusual features such as small genitals or wide spaced nipples. Very occasionally a health problem that emerges later such as epilepsy is the first sign.

Among 17 Unique families, 11 knew at birth that something was wrong. One had a pregnancy scan suggesting Down syndrome at 20 weeks. Among the others, delayed development was picked up between the ages of eight weeks and two years (Unique).

"In retrospect there was a lack of fetal movement towards the end of pregnancy. From birth Millie lost a lot of weight and failed to breast feed. At 3 weeks old Millie's grandmother noticed that one of her eyes wasn't moving in the same way as the other (she has Duane's syndrome). Her weight continued to be low and she was always late in hitting milestones. The health visitor was concerned from early on but convinced us that something was wrong when she failed to pass objects from hand to hand in her sixth month." 2q36.3 or q37.1 deletion

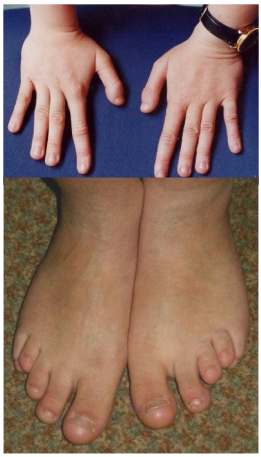
"Araylyn was born with a tracheoesophageal fistula which was discovered a few hours after birth because she was spitting up mucus and unable to clear it." 2q37.1 deletion

"Benthe was very, very quiet and weak after birth. She did not pick up drinking her bottles herself and was tube fed. She had some dysmorphic features and simian lines on both hands. She developed severe reflux." 2q37 deletion

Slight changes in facial appearance

Babies and children with a 2q37 deletion often have facial features that are similar to others with the deletion, and these features are still recognizable in adults. Typically, these include: a nose with small, swept-back nostrils and a V-shaped tip that hangs down a little; a thin upper lip and a small mouth; arched eyebrows; quite deeply set, narrow eyes, sometimes slanting slightly upwards; an altered ear shape or position; a rounded, bossed or prominent forehead; full cheeks or a round face; a flattened or short groove between the nose and the upper lip (known as the philtrum); occasionally, a small lower jaw; and, in some, thin or unruly hair. Most children have some, but not all of these typical facial features; children with very small interstitial deletions from 2q37 may have none; the features make some unrelated children look surprisingly similar (Falk & Casas 2007; Leroy 2012; Unique).

Hands and feet



The bones in the palms of the hands (metacarpals) are unusually short in around half of people with a 2q37 deletion. Sometimes the bones of the fingers (phalanges) are short too (brachydactyly), but in others the fingers look too long for their hands. When the hand is made into a fist, there can be a 'dimple' where the knuckle should be. You can sometimes also see this in the feet, with toes that are very short and oddly placed and feet that are often short and broad, making finding shoes difficult. The third, fourth and fifth fingers and toes are specifically affected, or sometimes just the fourth toes and fingers. This feature is not usually obvious in a baby, but becomes more evident with age. The short fingers and toes do not in themselves impact on a child's functioning.

There can be other unusual features, although these aren't specific to the 2q37 deletion syndrome: unusual spacing between the fingers and toes; narrow, tapering fingers; broad big toes; clubfoot (talipes) which may resolve with physiotherapy (Leroy 2012; Unique).

Flat feet and unusually flexible fingers are also very common (*see* **Joints**, page 22).

Pregnancy

Usually uneventful

Among Unique mothers, pregnancy was usually uneventful, although a few mothers noticed that their baby moved less than they expected. Two/20 developed diabetes in pregnancy, and diabetes has returned four years later in one case. One mother experienced bleeding. Babies with a 2q37 deletion are no more likely to be born early (Falk & Casas 2007). But in the Unique survey, 3/20 mothers had a raised blood pressure or pre-eclampsia, and six babies, almost one in three, were born prematurely, in the earliest case at 28 weeks (Unique).

Newborn

Most babies born in fairly good condition, but breathing and feeding problems common

At birth, most Unique babies were generally in a good condition. The Apgar score is a measure of general wellbeing at birth, and most Unique babies scored high, although those with breathing difficulties faced greater challenges.

Different babies: left, 2 months old; right, 6 months

Babies with a 2q37 deletion are no more likely than typically developing babies to be unusually small or big at birth (Falk & Casas 2007). Unique babies born around or after their due date were usually a good size at birth, weighing between 6lb 1oz/ 2.749kg and 9lb 11oz/ 4.4kg. Birth weights of babies born prematurely were of course lower, from 2lb 14oz/1.3kg to 6lb 6oz/ 2.9kg. Babies born to mothers with diabetes tend to be larger, and the two babies weighed 7lb 9oz/3.43kg and 9lb/4.082kg.

Some babies with a 2q37 deletion have initial breathing difficulties. In babies with a breakpoint at 2q37.1 this may be due to soft cartilage rings supporting the windpipe (tracheomalacia, *see* **Breathing, and childhood infections**, pages 23-24) or in babies with any breakpoint it may be due to soft, limp tissues in the back of the throat (laryngomalacia) (Casas 2004; Kitsiou-Tzeli 2007; Unique). 14/21 Unique babies had no breathing difficulties at birth, while seven did. Apart from one baby with a link between the windpipe and the food passage, corrected by surgery, the difficulties were not long lasting, but may mean that babies need extra help to establish their breathing in the first place, and extra oxygen for some days after birth.

A minority of babies are born with some visible birth defect, such as an unusually small head (Casas 2004); in boys, small genitals (Conrad 1995; Reddy 1999; Unique); clubfoot or talipes (Leroy 2012; Unique); or a cleft palate (the roof of the mouth is split) (Leroy 2012; Unique). Other babies, again a minority but a substantial one, are born with an important health problem such as a heart defect, or a structural problem of the digestive tract. *See* **Medical concerns**, pages 20-25.

Early hypotonia

Many babies are born with low muscle tone

Many babies - at least half - are born with low muscle tone, feeling unusually floppy to hold. Of 19 newborn Unique babies, 12 were very floppy, and this was more likely among those with larger 2q37.1 deletions than among those with smaller deletions. Tone tends to gradually improve and physiotherapy is helpful (Falk & Casas 2007; Unique).

Feeding and weight gain

Feeding and weight gain are often affected. Constipation is common

As a new baby, and at 8 years

Feeding difficulties affected 2/3 babies in the Unique survey, regardless of the size of their deletion. The typical low muscle tone can affect their ability to latch on, suck from the breast or bottle and to coordinate sucking with swallowing and breathing. Around 1/3-5 babies have a high palate (Falk & Casas 2007; Unique), which can make feeding harder. Other factors include your baby not crying to be fed; being very sleepy or tired by feeding; or if s/he has a health issue such as a heart or gastrointestinal problem. Rarer problems include choanal atresia, where the back of the nose is blocked by a membrane, so the baby has problems feeding from birth (Unique).

Feeds by bottle can take a very long time, and feeding from the breast may not always be possible, or babies may need very frequent feeds. Babies can take expressed milk by sipping from a spoon or cup or from an easy-suck nipple or bottle, but quite a few mothers are unable to maintain their milk supply in these difficult circumstances. Feeding problems tend to improve with time but in the meantime there are many ways to help a baby who is having difficulty feeding and, if necessary, it is possible to feed temporarily by nasogastric tube or through a gastrostomy tube direct into the stomach to ensure that a baby gets enough nutrients.

Gastro-oesophageal reflux is common. In babies with reflux (where milk flushes back from the stomach up the food pipe) there is a possibility that babies will inhale milk, putting them at risk of aspiration pneumonia. Careful feeding and positioning can help reflux as can feed thickeners and medication to inhibit gastric acid. Babies often grow out of reflux, especially when they start solids, although even on solids some children continue to bring back small amounts of food after meals. Reflux can be persistent, although most families can control it using prescription medication. If simple measures are not enough, it is possible to cure reflux with a surgical operation known as a fundoplication, in which the action of the valve between the food pipe and the stomach is improved.

Unique's experience is that many babies find it difficult to put on weight, known medically as 'failure to thrive'. They may need enriched or fortified milk and a high calorie diet once they move on to solid foods. Most babies put on weight satisfactorily once the initial feeding problems are sorted out, but a few have persisting weight concerns.

Babies may move late to solids, and some may benefit from fortified or adapted milks to meet their extra needs after six months. A wide range of finger foods and nutritious drinks is needed until children learn to handle a spoon.

The good news is that the great majority of babies eventually put their feeding problems behind them and by toddler age or soon after are eating with the rest of the family. Even children with the most severe feeding difficulties as babies do learn to eat and to enjoy their food, although, because of chewing problems, some need it puréed, mashed or cut up for longer than typically developing children.

Some children with 2q37 deletions have weak or extremely flexible hands. Some children

use cutlery when you would expect, but others only develop enough dexterity by the age of four or five. Families find two-handled cups, toddler or adapted cutlery and rimmed plates helpful. Children do eventually learn to feed themselves, although using a knife and fork together or a knife at all can be a special challenge. For some children food becomes a behavioural issue.

Some children have slow motility - the food travels slowly through the digestive system, due to their hypotonia.

Constipation is common because of low muscle tone, especially in younger children. Half the Unique children were affected at least as babies or toddlers, and it could be severe, so if the simple remedies of additional fluid, fibre and exercise are not successful, families should seek medical advice promptly. One family fed their child the 4 Ps - pears, plums, prunes or peaches, supplemented with a prescribed stool softener or laxative, others used massage, kiwi fruit, teas and herbal remedies. A small number of children had the opposite problem of long term diarrhoea and families controlled their child's fruit intake.

"Breastfed for 7 months. Very tricky at first but fine once we both learned what to do." 2q37.1 deletion

"Brooke had very narrow nasal passages so found it difficult to breathe and breastfeed. We used a humidifier to keep the air moist while feeding, and saline drops to keep her nostrils clear." 2q37.3 deletion

"Although I did not breastfeed JW, I pumped breast milk for his entire first year. We then went to goat milk and drinkable yogurts. He refused cows' milk except yogurt. I did have to add coconut oil so he could get the extra fat and nutrients." 2q37.1 deletion

"Millie lost a lot of weight and I was persuaded to put her on formula. She definitely managed to get some milk from me, but not enough. It may just have been that she was not hungry, as even on formula she failed to gain weight as expected, or a lack of muscle tone and coordination making breastfeeding hard for her, coupled with my inexperience. Her weight got up above the 0.4th centile at around age 3½." 2q36.3 or q37.1 deletion

"Benthe was tube fed most of the time. She seemed to be too tired to drink enough, delayed (weak) coordination (suck-swallow-breath), not hungry at all, never cried to be fed. Her weight gain was initially OK, but after 14 months she had a G (PEG) tube fitted, and a Mickey button after 20 months. When she was just 2, Benthe successfully joined a special 'hunger provocation' programme at a children's hospital in Holland. She was weaned from the gastrostomy tube in 3 weeks. Today, she eats thick liquids (vegetables, fruits, yoghurts). However, she is struggling to gain weight and does not eat enough yet to actually grow well." 2q37 deletion, 2 years 2 months

"Emma now eats everything we eat. She has three meals and 1-2 snacks a day." 2q37.1q37.3 deletion, 20 months

"I had trouble feeding Cody when he was first born: I tried to breast feed but he would not eat. We had to feed by pushing the teat on top of his tongue. Cody was put on some special milk which helped him gain weight. Before that, his weight was very worrying to myself and the doctor. After being on the milk, Cody got a big appetite and began to feed, but he still had trouble putting weight on. Today Cody eats everything. He has a very good appetite and will sometimes have 2 dinners at school. He loves gravy dinners and garlic dip. At the moment, it seems that he eats all the time and he doesn't know when to stop, so I am having to watch him closely." 2q37.3 deletion, 4 years

"Thom eats everything and anything and in excessive amounts." 2q37.3 deletion, 6 years

"Eats everything now." 2q37.2 deletion, 7 years

"Found acidophilus to be very helpful with preventing diarrhoea and good for his digestion and immune system." 2q37.1 deletion, 13 years

Tendency to overweight?

A possible tendency to overweight

There are many reports of children with a 2q37 deletion being overweight, and the tendency to overweight seems to increase with age, and can but doesn't necessarily progress to obesity (Casas 2004; Williams 2010; Leroy 2012; Morris 2012; Unique). As a result, doctors recommend families to encourage their child to be as active as possible and to watch their diet, but the typical low muscle tone can be a barrier to children taking exercise.

Unique's experience is more mixed: its 2003 survey showed that 50 per cent of families stated specifically that their child was thin or petite. Families at a 2q37 deletion meeting reported that some children under eight ate a large amount but had difficulty putting on weight, while other families reported overweight. The 2012 survey showed that it was three times as common for a child to have a normal weight than to be overweight, but many of these parents already knew about the potential problems of weight gain and watched their child's intake.

"We fear overweight to be an issue with a lot of other 2q37 kids, so we have not been very forthcoming when doctors wanted to put him on weight gain formulas. Had we done that he might be overweight now. We do not limit his eating or drinking, we just don't push super high calorie drinks/formulas." 2q37.1 deletion, 4 years

"Lilia likes soft, fatty foods, cream cheese, peanut butter, hot dogs, butter, toast, eggs and mayonnaise. She has just started thyroid medicines, which we expect to help control her weight." 2q37 deletion, 10 years

"Lucy is slightly overweight. We keep it under control with activity and limiting access to foods. Lucy is obsessed with food and eating and will steal foods if she can and would eat leftovers from the floor or bin." 2q37.1 deletion, 13 years

Growth

As a group, some children are short but individuals are most likely to be normal height or tall

Around 1/4-5 adults with a 2q37 deletion are somewhat shorter than you would expect for their family (Casas 2004; Leroy 2012). Children in the 2012 Unique survey were usually a normal height for their age; a few were tall and about twice as many were short.

Typically, both children and adults are not much shorter than you would expect and their body proportions are normal. As far as we know, there is no hormone deficiency; too few children with 2q37 deletions have tried growth hormone for the outcome to be known, and evidence from studies in other chromosome conditions is conflicting.

"Lucy is short - 136cm [4' 6"]. We are awaiting blood tests to check for growth hormones etc as her height has not increased for about a year now. As she started her periods 3 years ago she has probably stopped growing." 2q37.1 deletion, 13 years

Developmental delay

The great majority of children with a 2q37 deletion will experience some developmental delay. It is usually mild or moderate - so children function at around half their age - and

sometimes severe (Falk & Casas 2007; Leroy 2012; Unique). Considering all aspects of development, the profile is usually 'spiky', often sparing sociability and most obviously affecting mobility, although some children have normal motor skills. Very occasionally, delay is first obvious in a child's speech and language development.

"Millie is always behind but seems to get there eventually." 2q36.3 or q37.1 deletion, 4 years

"Melanie seems only a little less mature than some preschoolers her age." 2q37.3 deletion, 3½ years

Sitting, moving, walking (gross motor skills)

Children with a 2q37 deletion are often delayed in learning to sit and walk

Most often, babies hold their head steady and roll over late; they may never crawl or shuffle; they may need encouragement and sometimes physiotherapy before they learn to sit; they need support when they start walking and continue to need it for some months before launching out on their own. On average, Unique babies were able to hold their head steady between 6 weeks and 12 months; rolled between 3 and 14 months, although 2/22 babies never rolled. They sat up between 6 and 18 months and at first tended to slump. Those who bear-crawled, crawled or shuffled did so between 9 months and 2½ years. Most children took their first steps when they were 2 years old, though the range was 15 months to four years; they took a further 2 to 9 months to walk alone, and even then needed a pushchair when tired or unwell. Most children were 5 or more before they could climb stairs, but one or two mastered even this when they were 2. Actions like standing up from sitting on the floor remained a challenge for a few older children.

Once children are on the move, balance and fatigue remain ongoing issues. Children may lack a sense of danger and need help over uneven ground and soft surfaces. Their gait can be awkward or wobbly, with their feet apart, and some children give the appearance of limping, while others have a characteristic lope with their arms held out slightly behind them. They are likely to be late to learn other mobility skills like running, hopping and skipping. When running, they tend to hold their arms out 'as if on a rolling ship' to keep balance, but even so, they fall much more often than other children and need a safe play environment, especially since some don't have saving reflexes. A child who falls often and can't save himself may need a helmet. A few children have an uneven leg length – one leg is shorter than the other, so once wearing shoes they need an insole or build-up to correct the difference. In the children with persisting low muscle tone, walking is hard work and they tire easily. By the age of 10 or 11, children are usually walking quickly and well, but may still be clumsy.

Swimming is a helpful activity, as are well-supervised playground and soft play, dancing and riding for the disabled. Some children use a treadmill; other popular activities include trampolining, swings, multisports, playing chase, cycling (variable number of wheels), disability football and dancing. Despite initial delays, one 8-year-old was swimming, playing soccer, doing gymnastics, cycling and roller skating but this level of activity isn't possible for all. A 15-year-old was mastering a 2-wheeler and swimming a few strokes unaided (Unique).

Many children have very lax joints (*see* **Joints**, page 22). For example, the knees may curve backwards and the feet may be flat or angled so the child walks on the inside edge of the foot. Physiotherapy is usually key and and low impact physical activity may be helpful; most children will need an orthopaedic assessment and special supporting footwear, splints or insoles. Families try to ensure their child takes as much exercise as

Getting mobile, from top left: 19 months; 22 months; 2 years; 4 years; 9 years; 7 years (right).

possible to build up strength.

Congenital hip dislocation is a bit more common in babies and children with a 2q37 deletion than others (Casas 2004). This is screened for at birth but if the newborn screening was normal and concerns remain, a specialist orthopaedic assessment may be needed. Dislocated or dislocatable hips can be treated by splinting to hold the hips immobile in the best position for development; sometimes surgery is needed. A child who has been in an immobilising splint or cast for weeks may lose more muscle tone and will need intensive therapy afterwards to build up muscle tone and strength. Occasionally, families have reported other 'clicking', 'cracking' or dislocating joints, particularly knee caps (Unique).

"She loves to crawl and tries to pull herself to a stand. She loves to be in a stand to play with her toys. She likes to wiggle to music. She also likes to lay on the floor and wiggle her whole body. She likes doing somersaults, too. (But she's only allowed with mommy's help)." 2q37.1q37.3 deletion, 1¾ years

"Our gait specialist stated that her best therapy is the therapy she gets from normal daily activities and play. We also did some weeks of PT to help address the foot slap, though it is still unresolved." 2q37.3 deletion, 3¾ years

"She has hyperflexible joints in her ankles which makes walking hard, but it helps to walk whenever possible. Going up and down steps strengthens her knees. Walking on tiptoes strengthens her arches. Physiotherapy sessions are few and far between now, but she still gets special shoes through the physiotherapist." 2q36.3 or q37.1 deletion, 4 years

Development: co-ordination and dexterity (fine motor skills)

Low tone and flexible joints mean that children's hands can be weak

Low tone and flexible joints mean that children's hands can be weak. Babies may be late in learning skills such as using both hands together and picking up with thumb and index finger instead of using a fist. This impacts on play, on feeding themselves, on self care and on learning to make marks, scribble, draw and write; later on, on practical activities like cutting and opening containers. Children experience a range of difficulty, so some

have broadly age-appropriate skills while others are very delayed. In this situation, special needs toys – on loan or exchange, adapted cutlery, handled cups and, later on, easy-grip markers are all useful. Children may have additional problems – one Unique child has a tremor, so his hand has to be steadied. Occupational therapy is usually helpful (Unique).

"Doing really well at all fine motor skills and is pretty on point with other kids her age." 2q37.1 deletion,14 months

"Actually he has a rather strong grip and likes to play with very small objects and strings. He prefers to eat with his fingers but has been able to 'use' a fork for about a year. Still working on mastering this (stabbing food efficiently, etc). He is starting to like ball pits, and very much enjoys sensory play. For this we get a kids' size blow up pool, and use food (sugar, frozen fruit, whipped cream, pudding, cracker crumbs) - anything that is edible that won't harm him. He loves textures." 2q37.1 deletion, 4 years

"Two years of physiotherapy has helped Thom's leg and core strength. We continue ensuring he has a lot of exercise to try and increase and improve his strength – his shoulder girdle is weak and this continues to delay his hand writing. Thom loves the Wii and we believe that this helps his hand to eye coordination. He has extremely weak hands and fingers so this hugely delays his fine motor skills (zips and buttons) and is yet to master the pincer grip with mark making tools hence his delayed writing. To add insult to injury Thom is left handed and we have used a Stilo left handed pencil which does seem to help along with the rubber pencil grips." 2q37.3 deletion, 6 years

"Has trouble holding a pencil, still can't use a knife, she tends to only use one hand for everything." 2q37.2 deletion, 10 years

"Has no problems but would struggle with very tiny pieces." 2q37.1 deletion, 11 years

Self care

Daily tasks like getting dressed (especially socks) and undressed, washing and brushing teeth will also be difficult, although using stretchy clothes, Velcro fastenings and slip-on shoes will help. Children gradually progress with daily repetition, cues, prompts, reminders, laying out clothes, recognised routine and bribery but, as ever, some get further than others. As for toilet training, 13/19 children were dry and clean in the day time. The youngest toilet trained boy was 4½ years old; three girls of 3 years were trained. Of the six children not yet trained, five were boys, one of them 13 years old; the only girl was 3¾ years old. At night, two boys, aged 5½ and 7 years, were dry. One family mentioned how useful a chair potty is when training.

"Melanie can wash her hands, dress and undress, use the toilet and feed herself." 2q37.3 deletion, 3½ years

"Needs full care." 2q37.2 deletion, 3¾ years

"She dresses herself, brushes her teeth, combs her hair, and has just learned to take showers, but still needs some help with washing her hair. She makes her own lunch bag for school." 2q37 deletion, 12 years

"Leon can dress and undress with a lot of physical and verbal cues. Working on washing and brushing teeth. 'A bit' toilet trained, dry in the day with a schedule." 2q37.1 deletion, 12½ years

Remember it's slow but it comes when they are able.

Learning

Many children have a mild or moderate learning disability, but the spectrum ranges from very mild to severe

Children are very likely to need some support with their learning, although the extent varies widely. Evidence from 42 surveyed Unique families shows a scattered pattern, with great variation in the skills that young people achieved. Two children were considered to be learning at a level appropriate to their age; nine children were considered to have a mild disability, in 14 it was moderate and 17 families categorised the level of intellectual disability as severe.

One child of 3½ whose difficulties are so mild that she does not qualify for early intervention services can recognize and name many letters, is beginning to trace her name, can draw some shapes and can make an 'M'. She has no learning support in a regular preschool classroom.

Among those with a **mild** difficulty, one child is only 3-4 months behind her chronological age in her learning. Among those with a **moderate** difficulty, many of whom are learning at about half their actual age, a 12-year-old is progressing well with her English but has difficulty understanding abstract concepts and time and space.

Many children (almost 2 in 3 in the Unique 2012 survey) have difficulty concentrating, and this undermines their learning, so exercises to keep them on task and sensory breaks can be helpful, and some children benefit from medication to help them concentrate better. Many children have a better longterm than short term memory, and need frequent repetition. Music is a useful learning and reinforcing aid, and it is interesting that one 5-year-old is learning sounds and letters well through phonics. Children's learning strengths are quite scattered, but often include music and social activities, sometimes language, practical tasks and computer skills. Many children strongly wish to please, and this supports their learning. Parents put success in learning down to their child's determination and wish to please, response to reward systems, and their own persistence, especially with new things. They recommend integrated learning with a combination of group and 1:1 therapy sessions; visual prompts and timetables; social stories; and Applied Behavioural Analysis (ABA), an approach which uses scientific studies of behaviour to teach appropriate social behaviour through a system of rewards and consequences.

In terms of academic skills, Unique's 2012 survey shows that around half of the reading-age children can read, while half cannot. Among children aged 10 or more, again around half can read, half cannot, although how much children can read is very variable, from a 12-year-old reading and understanding books for children of her own age to a 13-year-old recognising shop names and logos. Four/19 children aged 6-12 can write, although even the oldest can sometimes be hard to decipher.

Among school age children, the great majority attend a special needs school or unit, or one designed for children with autism. One child attends a school for children with epilepsy. Four children attend a mainstream/ regular school. One family who switched their daughter to a special needs school commented 'Just wish we had sent her earlier'. Within the school, whatever type, only three children do not have full time or virtually full-time 1: 1 learning support, and only 2 children of school age do not have a statement of needs or education plan.

"Just be patient, your child will progress at his own speed. Find something that he loves and try to learn around that subject."

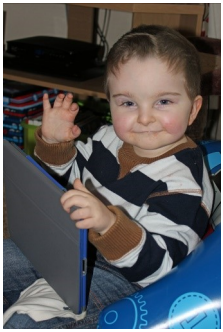
"Emma learns better when she is motivated. Food, music, and books are strong motivators. She works very hard and she is self-motivated. She likes praise, but would do her skills anyways. She likes learning new things, but becomes frustrated if she finds the skills too difficult. She would rather try her new skill on her own than with someone else. She is extremely independent." 2q37.1q37.3 deletion, 21 months

"Cody has a fantastic memory. If you tell him about something or somewhere we're going, he doesn't forget. If shown something on the computer, he can maneuver his way to it alone. At Christmas, he went several times onto different websites and searched for specific toys and pointed to them and the Christmas tree. When his grandparents asked Cody what he wanted for Christmas, he showed them. He loves anything computerized. He can use a Nintendo DS, WII and he now has an i-pad, which he loves and a Nabi tablet. He searches up Thomas the Tank and Super Mario on Youtube and e-bay. He is much brighter at using these than all of my friends' children of the same age. I am very proud of him." 2q37.3 deletion, 4 years

"His short term memory can be shocking: he can't remember what you've asked him to do ..... but anywhere he goes, even just once, he remembers where he went and who was there. He can recognise regular routes and tells us where we are, and where we might be going (shops/coffee shop/supermarkets). One of his strengths is PE, which we put down to the early years of physiotherapy. Also, information and computer technology, again in relation to his joy on the Wii. He is so very, very determined and motivated by sheer enjoyment at achieving a goal. Even if he's continuously last he considers he is winning as he is executing the mission." 2q37.3 deletion, 6 years

Speech and communication

Generally delayed, but most children learn to speak and some talk very well



4 years

Most children do learn to talk, but communication, speech and language are generally delayed, and any hearing loss adds to the delay. A small minority of children with a 2q37 deletion communicate primarily without words. Overall, the picture is quite varied and does not appear to relate to the size of the deletion.

The average age at which Unique babies first gave a social smile was 2½ months, about a month later than in a typically developing baby. The age range for the first smile was 1-5 months. Not all babies babbled (making speech-like noises), but those who did started from 9 months to 4 years. One baby was using words at 13 months, but lost her speech skills after she was taken out of a plaster cast to correct a hip dislocation. Once she started crawling, her language started to catch up again. The average age at which toddlers spoke their first words was almost 2-3 years (range 13 months to 6½ years) but vocabulary could remain limited and non-specific. 'Mama' can have many meanings! In one child, vocabulary dwindled after the age of 3 and by 12 years, he was communicating mostly with vocal noises and by pulling.

Yet other children as young as 5 or 6 are fluent speakers; a child of 3¾ speaks more like a 4-year-old. Generally, children speak better when they are well and fresh; only a few have any persisting difficulties in making the sounds of speech, and where speech sounds are unclear, they vary considerably from child to child. Usually, once any low tone in the face muscles has resolved, speech sounds become clearer.

As well as words, children communicate with eye contact, facial expressions, gestures, vocal noises, pushing, patting and pulling, humming, laughing and crying. They show a variable degree of success with signing systems, in part because low muscle tone can make signing difficult, and also because for some, signs can be hard to understand. For some, especially those with a hearing problem, signing unlocks the gates to communication, while others never master it and move directly to speech. A few use picture exchange systems or electronic aids. Some children use language repetitively, repeating the same question or phrase over and over again. Understanding is usually ahead of expression, but not in all children.

All families recommend speech therapy, starting early to develop eye contact through games, and working on shared attention; visual aids and timetables; one family recommends the Hanen programme (a programme aimed at promoting language, social and literacy skills (www.hanen.org)).

10 years

"Emma has some single words and a lot of babbling that we don't understand. She also says sounds for certain objects like 'ba' for ball and 'mmm' for milk. She will sometimes try to repeat what we say. For example, I told her that 'Mama was going to shower' and she repeated to me 'Mama shh'." 2q37.1q37.3 deletion, 21 months

"Millie talks fairly fluently but in short sentences and falls back on a relatively small number of stock phrases, often repeating things she has just heard. Her speech is not very clear." 2q37.1 deletion, 4 years 8 months

"Timaeus talks and signs; he uses fluent conversation. He learned to speak by mimicking and was very repetitive. It took a long time for understanding to catch up; we are still not sure whether he understands everything he says. Makaton signing really helped in the early days." 2q37.1 deletion, 5 years

"Thom communicates using fluent conversation. He understands if speech is broken down rather than bombarded – and forgets past a certain point – so he needs a large amount of repetition. We have been advised to keep sentences / instructions short and concise." 2q37.3 deletion, 6 years

" Lilia understands everything but cannot express fully how she feels. If she's sad, she can't say why. If hurt, she can't say what hurts." 2q37 deletion, 10 years

Two stories show how vital it is for the communication aspects of 2q37 deletions to be fully investigated and treated. A girl with a 2q37.3 deletion appeared developmentally delayed. In addition her parents noticed a lack of eye contact in babyhood. She was speaking some individual words at the age of four but by five her response to others' social or verbal overtures was limited. Testing at school showed a learning disability. At the age of 13, she learned to type with one finger. She can now type complex, grammatically correct sentences: something she never achieved through speech. By typing her answers she achieved a score of 107 on a verbal reasoning test and enrolled as a college student (Smith 2001).

Lucy, a Unique member, 13 years old, was provided with an iPad from her school. Her parents report that it 'has been amazing. She has surprised us and school staff at how well she can use it. She talks and she also uses the Proloquo2go program on the iPad'.

Behaviour

Families often find their children delightful but autism spectrum disorders are common

The Unique experience

When Unique asked families how their child behaved on a normal day, the great majority of answers were upbeat and encouraging. Families pointed to their child's cheerful disposition; and their sense of playfulness and humour, even as young as 2. Two reports in the medical literature also remarked on the child being 'very friendly' (Williams 2010).

"Delightful, cheerful, hilarious! Affectionate +++ "2q37.2 deletion, 3¾ years

More than two thirds of families in the 2012 survey said that their child was usually sociable and happy. Some take time to warm up, but typical comments are 'described by almost all of her doctors as 'the happiest baby they have ever met' (21 months); 'very tactile and affectionate, giggly, bubbly' (3¾ years); 'very sociable, popular and cheerful' (5 years); 'friends of all ages, some with special needs and others not' (6 years); 'loves people, likes to ask questions and be around friends' (10 years). A few children, especially younger ones, are more relaxed with adults; some have difficulty being understood by other children; others lack social boundaries and risk being too friendly, so they invade other children's space, or have no stranger awareness. Older children may find the intricacies of pre-teen friendships hard to negotiate as they slip, sometimes imperceptibly, behind in maturity and subtlety while retaining a strong, naive urge to be friendly. Families and regular carers are very important to them.

Two differing pictures:

"I've become a childminder and have 13 children on a weekly basis which Thom loves. He is extremely friendly, too trusting, which scares us, but is very popular at school." 2q37.3 deletion, 6 years

"Chloe finds friendships very difficult. She is very immature compared to her peers, becomes obsessive w ith friends which annoys them, and becomes overbearing." 2q37.3 deletion, 11 years

11 years

Among the possible problems that families mentioned were passivity (2 years); mood swings and frustration (4 years, 7 years); biting (4 years, 5 years); attention seeking (4 years); yelling or screaming; tooth grinding; anxiety (5 years); emotional immaturity (6 years); aggression when out of comfort zone (7 years); temper tantrums (4 families, including 2 children over 9); loudness (11 years; 13 years); generally challenging behaviour (13 years). Many aspects of behaviour in children with 2q37 deletion syndrome are well known to any parent of a small child. However, the behaviours are more intense, more extreme, they last longer and parents need extra ingenuity and energy to cope with them.

Here are some examples, with families' solutions:

"Getting overexcited. Rather than go downstairs, she will throw herself down" - 2 years
"Ignoring instructions, continuing to do what she wants. What works? Trying to be consistent" - 4 years

"Spitting on anything shiny or windows. Time out has reduced this"- 4 years

"Grabbing other children in his excitement. Distraction worked, as telling him not to had the opposite effect and he enjoyed the attention" - 6 years

"She gets frustrated when she can't have things. Keep explanations simple, use time out for tantrums, lie if necessary!" - 8 years

"Can show temper and lash out unpredictably as well as getting frightened by the unknown. Now his speech has improved he can be reasoned with when having a tantrum or, even better, defused by distraction" - 10 years

"Extreme anxiety. Fluoxetine" - 10 years

Among other management techniques mentioned are music, social stories, reward systems, praise and, more formally, Applied Behaviour Analysis (ABA). One child, already on the maximum dose of risperidone for his age, was referred for specialist support with challenging behaviour.

Autistic behaviour

Many reports in the medical literature have concluded that the typical behaviour of a child with a 2q37 deletion overlaps that of a child with an autism spectrum disorder (Falk and Casas 2007; Williams 2010). Half the children in the 2012 Unique survey showed autistic traits, but only half of these were diagnosed with an autism spectrum disorder. Families often say that autism undermines their child's development more than other symptoms (Unique).

People with autism spectrum disorders have difficulties with social interaction (such as poor use of eye contact, facial expression, gesture); communication (delayed / impaired speech without attempts to compensate by signs or gesture, poor understanding of others' emotions, not seeking to share enjoyment, such as by pointing things out, bringing things to show); idiosyncratic use of words or phrases; rituals / repetitive behaviours (such as finger twirling, adherence to routines); restricted interests and poor imaginative or imitative play.

Here are some families' experiences:

"Handflapping, humming, stimming. Tends to play with things (toys) in the same way all the time." 2q37 deletion, 2 years 2 months

"We noticed the limited eye contact and social interactions at 9 months. Eye contact has improved but is still limited. Rocking has reduced as she develops more ways to play and now she just appears to rock in situations she is unsure of" - 4 years

"The school wants Millie to be tested for autism with a view to reapplying for a statement of special educational needs. She has some behaviour which might be autistic, or might be due to developmental delay: she sometimes laughs when people are hurt; she is not very interested in imaginative play; she doesn't understand when her little sister is pretending to cry as part of her imaginative play and so she gets upset; she fixates on certain book and TV characters and gets very upset eg if not allowed to hold her character, doll or picture." 2q37.1 deletion, 4 years 8 months

"She is very repetitive, however she is also very social and likes to be touched" - 5 years

"Self rocking movement for stimulation, staring at lights, rocking himself to sleep at night, won't sleep without a pillow over his head, poor social behaviour, though getting better as he gets older, likes routine, uncomfortable in new environments or with new people around him." 2q37.1 deletion, 7 years

Two features stand out:

Odd obsessions

Two thirds of families in the Unique 2012 survey remarked on their children having an odd obsession. Children can become obsessed with anything - a texture; an activity, a routine, an object. Older children can become very knowledgeable about their obsession,

or skilled with it, and for younger children it can be used as a reward. A child will carry the object of his obsession with him at all times, even taking it to bed. Children's obsessions include: looking at and playing with their fingers; the weather; hair, feathers, newspapers, certain toys; goggles, cars, shoes, rubbery textures; vacuum cleaner; strings, doors, rings; pirates; the Wii; racing cars; plugs, drains; catalogues and leaflets; magazine pictures.

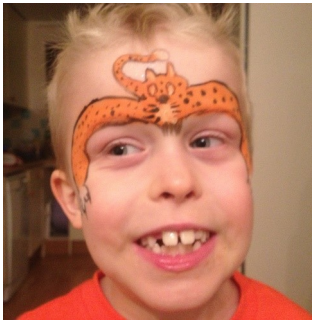
Repetitive behaviour

Two thirds of families in the Unique 2012 survey told us that their child had some form of repetitive behaviour. This was often speech, either repeating heard sounds and phrases again and again or more often asking the same questions (Is the sun going up or down? What are we doing tomorrow? Where are we going?) repeatedly until deliberately stalled. Younger children were more likely to repeat a simple action, like opening and closing doors; turning light switches on and off; taking shoes on and off; banging. A few children appeared to repeat an action for comfort, such as rocking. Many played their games always in the same way. A typical example:

"Marco wants repetition of the same activity or task. If he likes what he is doing, he will continue to prompt for it until he becomes exhausted. For example, running up and down the hallway, pushing a music key on a game over and over again, throwing all the items out of the bathtub." 2q37.1 deletion, 7 years

What do children with a 2q37 deletion enjoy doing?

Under 3 Anything with music. Mickey Mouse, Cookie Monster, Elmo. Mirrors, dolls, putting things in and taking things out of containers. Playing with her cat and dog. Playing with her family. She loves sitting with you and reading a story. Bumba, Miffy the Bunny, iPhone, watching TV, cuddling, bathing, romping, playing peekaboo, moving around on her small bike, exploring, making a mess, playing on her little computer.



6 years

3-4 years Playing with her siblings, baby dolls and other toys. Also likes to play outside and watch TV. Had a recent interest in colouring. Shoes. Ring type toys, banging things, iGoTo Jungle app on iPhone, taking a ring or plate object and hiding it or throwing it and then going to get it. Anything to do with Thomas the Tank Engine, Super Mario, Nintendo 3DS, Nintendo Wii, iPad, nabi tablet and books. Watching his videos; just starting to collect things to do with Ben 10. TV, her favourite DVDs, and books.

5-6 years Pirate toys, role play, films - anything to do with pirates especially Jack Sparrow & Pirates of the Caribbean. Castles, knights and star wars. Cats, music and cartoons. The Wii, as yet only very young games, very good at Donkey Kong and Mario games. TV shows such as 'Wipeout' ; older movies like Wizard of Oz and Charlie and the Chocolate Factory. Madagascar movies and now Batman. Toys & CBeebies on TV.

7-10 years iPad, music, chasing our 2 kittens, trampoline. Swinging at the park, cycling, music, watching children's TV. Computers, music DVDs. People, pets, pretend play kitchen and doctor, stuffed animals.

11-13 years Laptop, dolls and buggy, magazines, music player. Pets. Playing instruments (piano and cello), playing with other people, TV and videogames. Magazines, catalogues and leaflets. Soft toys and cars, which she likes to line up. Playing on her iPad – particularly drawing games. Colouring with crayons.

Sleep

Around half of children with a 2q37 deletion have a sleep problem

2q37 deletions can be associated with sleep problems (Falk and Casas 2007; Williams 2010). Unique's experience is that around half of the children with a 2q37 deletion have sleep problems; the others do not. The most common persisting difficulty is night waking and being unable to settle back to sleep. One child had sleep apnoea (spells where he stopped breathing during sleep) as a baby resulting in disrupted sleep for all. Having his adenoids and tonsils out has helped, but the waking habit persists. Families have tried many remedies, including, in the UK, the Cerebra sleep service for special needs children [www.cerebra.org.uk]; bedsharing; melatonin and other medications; blackout curtains; using a clock. Children over 7 or so do learn to settle themselves back to sleep or at least to stay quietly in their own room. One older child wakes complaining of aching legs and anxieties.

The burden of a persistently waking child is enormous, and families should all have access to a sleep service, ideally designed for children with special needs.

Medical concerns

Two thirds of babies are born without any major birth defect

Heart

Around one baby in 5-7 with a 2q37 deletion is born with a heart problem (Falk & Casas 2007), although in one recent series of 14 children and adults, no-one had a heart problem (Leroy 2012). Some heart problems are serious and may need surgical correction; Unique's experience is that many problems are identified but resolve naturally without treatment. Because of the frequency of heart problems, children with a 2q37 deletion are recommended to have an echocardiogram and cardiac assessment at diagnosis. In the Unique series, all children are now thriving after outgrowing the problem or after surgery (Unique).

The commonest heart problems are septal defects (holes in the walls between the two sides of the heart) (Casas 2004). In an atrial septal defect, the hole or holes are in the barrier between the two collecting chambers, while in ventricular septal defects they are in the barrier between the pumping chambers. The holes allow blood to pass from the left of the heart where the pressure is higher, to the right side. Babies are not blue, but this puts extra blood flow through their lungs and makes the heart work harder and children need to be seen by a cardiologist. Some small holes close up naturally, but if they do not close, or they are very big, children need surgery.

Another problem is a narrowing (coarctation) or poor growth (hypoplasia) of the aorta near where it comes out of the heart, which reduces the blood flow to the rest of the body; this too needs surgical treatment.

Other heart abnormalities have been found, including persistent ductus arteriosus and patent foramen ovale (both persisting structures of the fetal heart); and a bicuspid aortic valve (the valve that regulates blood flow from the left ventricle into the aorta has only two instead of three flaps or valves) (Falk & Casas 2007; Unique).

Kidneys

Kidney anomalies occur in around 1:9 people with 2q37 deletion, but possibly more as many have no effect on health and are only found on screening (Falk & Casas 2007). Unique's experience is that 4/22 children had something unusual about their kidneys, but

none had any problems caused by this. Two children had horseshoe or U-shaped kidneys (the kidneys are joined by a bridge of tissue); one had a kidney in the wrong place (ectopic). Other kidney abnormalities include: a small kidney on one side that doesn't function well; this can cause progressive damage and high blood pressure so the child needs monitoring; or one kidney can be on the wrong side and joined to the other kidney. So-called 'duplex' kidneys with two sets of ureters (tubes leading to the bladder) can occur, causing backflow of urine up the ureters and kidney infection. An ultrasound scan should identify any of these kidney anomalies and if found the child should be seen by a kidney specialist.

There is a suggestion that children may be more likely to develop kidney cysts as they get older, based on two children in the medical literature who had normal kidney scans in the first two years of life but were found to have kidney cysts later. No Unique children had kidney cysts. Kidney cysts are quite common in adults and most do not need treatment or cause problems. They are not cancerous. Occasionally the cysts can cause pain or be associated with blood or infection in the urine for which more detailed investigations may be necessary. Because of this, it is currently recommended children have kidney ultrasound scans at diagnosis, at 4 years and at around 15 years; and further testing if they are having urinary tract infections (Falk & Casas 2007).

Wilms tumour is a type of kidney cancer that can occur in children, mostly under five years, so a regular (3/6 monthly) ultrasound scan of the renal and urinary system is usually offered until the age of eight. Of more than 100 cases of 2q37 deletions published in the medical literature, three children have developed Wilms tumour, all at a very early age. Two of the children had large deletions including 2q37.1 and the other child had an extra risk factor for Wilms tumour. There are so few cases that we don't really know what the risks are, but Wilms tumour appears to be uncommon among those with a 2q37 deletion and has not been seen in Unique's 87 members. The available evidence suggests that children with larger deletions may have a slightly higher risk. Most Wilms tumours respond well to treatment and most children can be cured (Falk and Casas 2007; Aldred personal communication; Unique).

Hernias

Various types of hernia have been found, including diaphragmatic hernias (there is a hole in the muscular sheet separating the abdomen from the chest, allowing the contents of the abdomen to gather in the chest and take up space needed by the growing lungs). These are usually picked up on the 20-week pregnancy scan and dealt with soon after birth. Inguinal hernias (part of the intestine from the abdomen bulges into a sac near the scrotum or the vagina) and umbilical hernias (part of the abdomen bulges out through the umbilical ring at the navel) are fairly common. Umbilical hernias can be minor and self-heal in time. However, diaphragmatic hernias and inguinal hernias need surgical correction. Unique's 2012 survey showed that 1:3 children, all but one with a definite 2q37.1 deletion, had some sort of hernia, and one child had umbilical and inguinal hernias and incorrect attachment of the diaphragm, all needing surgical repair (Casas 2004; Falk & Casas 2007; Unique).

Gastrointestinal abnormalities

The medical literature points out a number of anomalies of the digestive tract that can occur with a 2q37 deletion. Unique's experience is that these are not common, with just 3/88 members affected. Generally all need surgical repair.

The intestine can twist on itself. It should be anchored at various points but if the small intestine and part of the large intestine isn't anchored properly (malrotation), it can twist (volvulus). This is a surgical emergency and may present with episodes of vomiting, disproportionate abdominal pain or red stools. Other problems include narrowing or blockages at any point in the gastrointestinal tract. A gastrointestinal blockage is a surgical emergency. Additionally, pyloric stenosis has been seen, where the ring of muscle at the outlet of the stomach becomes thickened and narrowed, stopping food leaving the stomach and causing forceful vomiting at a few weeks of age (Falk & Casas 2007; Unique).

Apart from two children with malrotation, one of whom presented with volvulus, and one with a tracheo-oesophageal fistula, problems seen in Unique were not structural. They include one child with coeliac and Crohn's disease, requiring an ileostomy (a surgical procedure to link the end of the small intestine to an opening in the abdomen); two children who had a rectal prolapse (the lining or entire wall of the rectum protrudes through the anus) associated with either diarrhoea or constipation; and a large number of children with constipation (*see* **Feeding**, pages 8-10).

Brain

Evidence from imaging children's brains has shown that slightly enlarged ventricles may occur. There have also been isolated instances of hydrocephalus, where the fluid-filled parts of the brain are enlarged and pressure within the brain may be increased; holoprosencephaly where the two halves of the brain haven't separated properly; and problems with the cerebellum that mainly controls balance and coordination (Casas 2004).

Within Unique, three children had hydrocephalus, and one child has shown enlarged ventricles, but these have reduced as he has grown. Another child has a somewhat small cerebellum; two further children have non-specific changes on brain scan. Two have craniosynostosis (early fusion of some of the bones in the skull, so that the brain is cramped as it grows) which was corrected by surgery (Williams 2010; Unique).

Joints

Sitting in a W-shape

Loose, overly flexible ('bendy'), or clicky joints are common in all young children but even more common in children with a 2q37 deletion. Only 4/19Unique families surveyed in 2012 said their child *didn't* have unusually bendy joints. Virtually any joints can be affected: hips, knees, ankles, wrists, fingers were mentioned most often. The loose joints are partly a result of low muscle tone but may persist even when overall tone improves. As a result, children may adopt an unusual posture: sitting in a W-shape; and slumping. They may have a 'rubbery' walk, with their knees bent backwards and flat feet. They may find gripping and grasping especially difficult. Most children need physiotherapy and some need supports in their shoes to realign the feet.

Joints are usually not so loose that they can actually dislocate, although this can happen. When hypotonia occurs before birth, the sockets may not form deeply enough, leaving joints prone to dislocate. One Unique child spent 4 months in a plaster cast to correct her hip sockets. Another girl was found to have an underdeveloped shoulder joint (Wilson 1995; Unique).

Seizures

Reports in the medical literature show that seizures affect 20 to 35 per cent of children with a 2q37 deletion, but this total includes children who only experienced febrile convulsions. In some babies, complex febrile convulsions progress to frank epilepsy. Seizures were more frequent among Unique members in the 2012 survey, affecting almost half. Children with seizures usually had no structural problems in the brain and responded well to standard anti-epilepsy treatment (Aldred 2004; Falk & Casas 2007; Unique).

Seizures are not usually seen in very young babies, although one Unique baby also had apnoea (spells where he stopped breathing), which confused the diagnosis. Various types of seizures occur, including absences, tonic-clonic and myoclonic seizures. They tend to respond well to standard anti-epilepsy medication, although a few children have severe side effects from medication and seizures that are very hard to control. Parents note that seizures are worse and more frequent when their child is unwell. One child has had vagal nerve stimulation (VNS), where a battery-powered device similar to a pacemaker is implanted under the skin delivering mild electrical stimulation to the brain via the vagus nerve, but it is too soon to judge its success.

Genitals

Up to six per cent of children have a genital anomaly according to medical reports, although the rate is much higher in Unique, almost 50 per cent of boys in the 2012 survey. In boys, undescended testes, a common problem regardless of chromosomes, may need to be brought down surgically; the testes may be small and poorly functioning; hypospadias, where the outlet of the tube from the bladder (urethra) is on the underside of the penis, may need surgical correction if it's significant; the penis itself may be small (Falk & Casas 2007). One Unique boy with hypospadias and undescended testicles has a low level of testosterone. In girls there is one report of a divided uterus and underdeveloped ovaries (Viot-Szoboszlai 1998); another has a forward location of the anus; a boy has a slight web of tissue between the penis and scrotum (Unique).

Spinal curvature

Spinal curvature may be more common than in typically developing children, due more to low muscle tone and muscle imbalance than to vertebral abnormalities. It was seen in 9/20 children in the 2012 Unique survey. It may need monitoring, bracing or surgery. Scoliosis (a sideways curve), kyphosis (a backward curve, creating a hump) and lordosis (an inward curve) may all occur. Among Unique children, none needed treatment until around the age of 11, when two children needed physiotherapy (Unique). When Unique reviewed spinal curves in adolescents and adults we found no further deterioration.

Bones

Osteopenia (slightly decreased bone mineral density) may also occur and is treated with calcium supplements and an increased vitamin D intake. One Unique child has been diagnosed with rickets (softened, weakened bones caused by lack of vitamin D, calcium or phosphate) (Unique).

Breathing, and childhood infections

Children with a 2q37 deletion generally have more breathing difficulties than other babies. A few have spells where they stop breathing (apnoea), but much more common are breathing difficulties caused by respiratory infections. Unique's records show that

half the children had repeated and troublesome chest infections including bronchiolitis and pneumonia that often needed hospital treatment. In one child these problems were eliminated by having his tonsils and adenoids out, but 2 children have had such frequent and severe chest infections that their lungs are scarred, and one child has bronchiectasis (the airways are widened, excess mucus builds up and the lungs are left prone to infection). Some children also have asthma, which may be triggered by viral illness and was treated with inhaled treatments. However, we have no evidence that this is more common than in typically developing children (Unique).

Babies, particularly those with a deletion including 2q37.1, are at risk of tracheomalacia, where the cartilage rings that keep the windpipe open are too soft, so the airway can close off more than it should. This usually causes noisy breathing that is worse when your baby is crying, feeding or has a cold and doesn't respond to standard asthma treatments. A baby who has tracheomalacia who has an infection may need hospital treatment or ventilation. Tracheomalacia gradually improves with age and is usually not a problem after the age of 2.

Eczema

Research reports suggest that one baby in three or four has eczema and that the eczema can be quite severe, although it does respond to standard treatments. Among Unique children, eczema is more common, affecting almost half the children surveyed in 2012. In one child, it was so severe that it was a diagnostic sign of the 2q37 deletion. Other children had it more mildly, generally responding well to standard treatment. It improved with age, so that by the age of 13, it was only slight or occasional (Falk & Casas 2007; Unique).

General wellbeing

Most families told Unique that their child was generally healthy, often much healthier than when they were younger and had frequent ear or chest infections. Some commented that their child with a 2q37 deletion was unwell more often than their other children, and more than 1/3 had been admitted to hospital at some stage with an infection-related illness. One four-year-old was in hospital more than 40 times.

Other than infections, illnesses seen in Unique's group of children with a 2q37 deletion include: possible abdominal migraines; iron deficiency anaemia, also seen with enlarged spleen (Polityko 2004); Bethlem myopathy, which may be associated with mutations of a collagen gene (*COL6A3*) gene in the 2q37.3 region; coeliac and Crohn's disease. One child had unusual thyroid test results, but without evidence of illness; two others had hypothyroidism (low thyroid levels) (Williams 2010).

Five children had intolerances or allergies, but this is no higher than in a group of typically developing children. They include lactose intolerance; milk allergy as a baby; dairy and cat allergies and asthma; hayfever; pollen allergy; a 'very mild and improving' allergy to strawberries, kiwi and fish.

Hearing

Some children have a degree of hearing loss, but the temporary type of hearing loss caused by glue ear is very much more common than a permanent hearing impairment. Two/21 children in the Unique series have moderate to severe permanent hearing loss and need hearing aids. Children can be significantly handicapped by glue ear, so regular checks and prompt intervention are recommended as needed.

Eyesight

Structural problems of the eyes are rare, but it is recommended that common problems such as a squint (strabismus), a refractive error (long or short sight), lazy eye (amblyopia, a healthy eye with poor vision usually caused by squint or a refractive error); astigmatism and ptosis (a droopy upper eyelid) are looked for and corrected if necessary. Within the Unique families, one child has nystagmus and difficulty synchronising the movements of his eyes; another child has Duane anomaly (a defect in the way the eye moves); in a further child there is concern over narrowing of the canal through which the nerves from the eye pass through the skull to the brain.

Teeth

In Unique's experience, children with a chromosome disorder generally have a higher rate of dental problems than typically-developing children. This may be due to a number of problems: unusual dental development; unusual size of the jaws and shape of the palate, leading to overcrowding or widely spaced teeth; feeding difficulties and delayed eating and chewing activity; tooth grinding, wearing down the enamel; poor enamel formation; unavoidable side effects of necessary prescribed medications; dislike of tooth brushing and going to the dentist. Teeth may emerge late and milk teeth may be late to fall out. Extra teeth may be found and either milk or adult teeth may be missing. Unique families of a child with a 2q37 deletion have drawn attention to other problems: damaged or chipped teeth caused by falls; chipped teeth where the cause is unknown; and fragile, easily broken and crooked teeth. The unusual combination of potential problems means that children may need sensitive and specialist dental care.

Different girls growing up: from left, 10 years, 10 years, 11 years, 13 years.

Puberty

Puberty starts at the normal age (Falk & Casas 2007), but there is one report of delayed puberty (Kitzio-Tzeli 2007). 'Normal' means 8-14 years in girls, with periods starting on average at 11 years; and 9-14 years in boys, on average 12 years. Unique's experience confirms this observation, with girls entering puberty from 9-12 years and starting their periods at 10-12 years. Behaviour can worsen with puberty (Unique).

Outlook

Children without major malformations and autistic features are currently believed to be those most likely to look forward to a long and healthy life. The oldest people with a 2q37 deletion reported in the medical literature are in their 60s (Syrrou 2002; Villavicienco-Lorini 2013) and Unique's oldest member is in his forties. Severe heart problems, major neurological malformations, severe, uncontrollable epilepsy, Wilms tumour and, perhaps, progressive cystic kidney disease may compromise life expectancy (Falk & Casas 2007). *See* also Unique's guide to **2q37 deletions in adults & adolescents**

Management recommendations

At diagnosis:

Echocardiogram or assessment to check the heart (all)
Kidney ultrasound, repeated at four years and around puberty (all)
Baseline eye evaluation
Consider hip evaluation if significant hypotonia
Hearing screen

Surveillance in babies and young children:

Weight: early feeding problems
Early developmental assessment & appropriate intervention
Check that newborn hearing test was normal. Periodic hearing tests for middle ear dysfunction
Ultrasound scan of the kidneys at four years to look for cysts
Screening for Wilms tumour in children with deletions that include band 2q37.1.
Babies and children should have 3-4 monthly ultrasound scans until age 5-7 years.

Surveillance in older children:

Height and weight: prevent obesity
Ongoing developmental assessments with appropriate intervention and statement of special educational needs/IEP
Periodic hearing and sight tests
Repeat ultrasound scan at 15 years to look for renal cysts
Early investigation of any abnormal symptoms
Behavioural assessment as appropriate

Why did the 2q37 deletion occur? Did my baby get it from me?

2q37 deletions usually occur out of the blue for no obvious reason. Less often, they can be inherited via the mother or the father. The only way to be certain is to check the chromosomes of both parents.

If both parents have normal chromosomes, the 2q37 deletion is a new occurrence. The genetic term for this is de novo (dn). A new 2q37 deletion has been caused by a mistake either when the parents' sperm or egg cells were formed or in the very earliest days after fertilisation. As a parent there is nothing you could have done to change or control this. Babies with pure 2q37 deletions are usually born to parents without any relevant chromosome changes. Even having the normal variant at the tip of the chromosome (*see* A normal variant in the general population, page 4) makes no difference. But a parent with a deletion that is only very slightly bigger than the normal variant can pass it on. A man reported in the medical literature passed a very tiny 2q37 deletion on to his daughter; he was unaffected by the deletion, but his daughter was affected (van Karnebeek 2002). A tiny deletion in 2q37.3 was found in three generations of another family (Villavicienco-Lorini 2013). A Unique child is reported to have inherited her deletion and duplication, both within 2q37.3, from her biological mother.

In a few families, one parent has a structural rearrangement of their own chromosomes. This is usually balanced so that all the genes and chromosome material are present and the parent is entirely healthy. However, these families have an increased risk of having another affected child (*see* page 3, Genetic testing *example 3*).

Whether the deletion is inherited or *de novo*, there is nothing you did as a parent to cause the 2q37 deletion. No environmental, dietary, workplace or lifestyle factors are known to cause these chromosome changes.

Can it happen again?

In families where both parents have been tested and have normal chromosomes, the risk of having another child with a 2q37 deletion is minimally higher than anyone else's risk. If a blood test shows that either parent has a balanced chromosome rearrangement involving 2q37, the possibility of having other affected pregnancies rises considerably. Someone who themselves has a deletion of 2q37 has a possibility of passing it on of about 50 per cent in each pregnancy and a 50 percent chance of a pregnancy with normal chromosome 2s.

Your genetics centre should be able to offer counselling before you have another pregnancy and if you already have a child with the 2q37 deletion, prenatal diagnosis will be possible if that is what you choose.

Can my child have children of their own?

It isn't yet known whether the condition affects fertility, but most girls with 2q37 deletions generally seem to go into puberty at the expected age and have periods normally which suggests they are likely to be fertile. For boys and men, there have been several reports of physical differences such as hypospadias, undescended testes, and small or abnormal (dysgenetic) testes which might be expected to reduce fertility (Falk & Casas 2007).

Anyone with a 2q37 deletion has around a 1:2 (50 per cent) chance in each pregnancy of passing it on.

Genes

There are 197-plus genes in the 2q37 region, but we don't know yet what most of them do. Until recently, 11 of them had been suggested as possibly involved in the 2q37 deletion syndrome but recently, a team of French researchers has highlighted more 'candidate' genes for various aspects of the syndrome (Leroy 2012).

While identifying the gene(s) responsible for certain features of 2q37 deletion syndrome is valuable and may help guide future studies, it does not lead directly to immediate improved treatment. Also, even if the supposedly responsible gene is missing, it does not always mean that the associated feature(s) will be present. Other genetic and environmental factors often have a role in determining the presence or absence of a particular feature.

The one gene that is fairly securely linked with many aspects of the syndrome is known as *HDAC4*. *HDAC4* is a gene in 2q37.3, whose genomic location is designated as 239,969,864-240,323,348 (*see* diagram on page 3). It is critical for proper bone and cartilage development and proper development of the heart. It also acts in the survival of nerve cells and plays a role in the development of behaviour disorders, in seizures and in intellectual disability. In the past it has been linked with the typical shortening of the bones in the hands and feet, but some people have lost this gene and have perfectly normal hands and feet, so more remains to be discovered. It seems likely that a chromosome disorder that interferes with the expression of this gene, even if it doesn't delete it entirely, can also cause mild symptoms of the 2q37 deletion syndrome. But the factors that interfere with *HDAC4* being properly expressed have still not been fully explained. Perplexingly, some people can be missing *HDAC4* (and other 'candidate' genes) but have none or only some of the expected symptoms (Williams 2010; Leroy 2012; Morris 2012; Villavicienco-Lorini 2013).

"Emma seems to be doing more than her doctors originally told us she would be doing at this age. I no longer focus on the 93 genes that she's missing, but on the 20,000+ others that are making her extraordinary."

What is special about your child?

"The good times far outweigh the bad times. She is delightful, has made us learn to sign, and communicate better."

14 months: "She is amazing. She has opened my eyes to so much more than I ever noticed before. Although for her sake, I wish she did not have to go to the doctors so much, I have learned a ton and learned to appreciate every moment!"

19 months: "She's taught me that there are more pathways in development than just what typical children do. Because of her, I have become a better teacher and mother. I am more patient and give my students multiple ways to show success. I have learned that the brain can do extraordinary things when given the opportunity. I make sure Emma experiences something new every day. I know it may sound strange, but she's also taught me to look up. We all look down at the ground so often we don't even notice anymore. I used to watch her look up at the sky, or up at lights and wonder why she did that. One day I decided to look up too, and it was beautiful. I truly believe she sees the beauty in everything and I want to be just like her. I'm excited to spend my life with her, no matter what her future holds. I no longer want her to be like everyone else... I wish others were more like her."

26 months: "Benthe has taught me to appreciate life, and take every day as it comes. She taught me that it's the way you deal with problems, not the problems themselves themselves that shape your personality.. "

3½ years: "She is an extremely loving, friendly and happy girl. She makes me smile."

4 years: "His smile and laugh are intoxicating. He has given me more strength than I knew I could ever have and more joy than I have 4 years: Cody is a special little boy and everything he does makes me proud, I love him more than anything, he makes me so happy. He is so loved by all our family."

4 years: "She has a gorgeous smile and a wicked laugh. Her enthusiasm for the things she likes makes her really enjoy life. She is affectionate and funny and sometimes wise. She makes me appreciate that there are more important things than being clever."

5 years: "He is so cheerful and loving, he brightens my day."

6 years: "He makes me laugh, has the greatest sense of humour, and is beautiful to look at. He is the most determined person I've known; whatever the challenge, he is courageous and just jumps straight in and has a go."

6 years: "An amazing, determined wee girl. Despite all the health issues and difficulties she keeps on going."

7 years: "His tenacity, he tries so hard, he loves life. His laugh and his smile."

11 years: "Lovely natured, kind and loving."

12 years: "She is extraordinarily happy, smiling, warm and caring. She has an unexplainably positive attitude and turns sad and disappointing things into something good, she truly looks on the bright side of life. She smiles and laughs a lot and wants to heal pain and hurt around her, with some comforting words or a hug. She feels and cares deeply for her animals. She is tidy and orderly and has a great aesthetic sense, appreciates nice clothes, colours, jewelry and music. She has a great singing voice and very good pitch. She plays the cello beautifully and composes her own songs on the piano."

12½ years: "Loves people and is very thankful for attention."

13 years: "Lucy has a great sense of humour and makes us laugh. She has recently learned to wave and will wave madly when she leaves for school on the minibus. She has recently learned to say 'I love you Mummy/Daddy/Robert/Charlie' using her iPad and ask for a hug. She taught us about disability and introduced us to some amazing people we would never have met."

19 months

3½ years

4 years

9 years

A gift for music?

Many parents have commented on their child's unexpected talent for music. A few children play musical instruments, others have a good singing voice, for others music therapy is important. Individual achievements depend on many factors such as hearing, fine motor control and the ability to concentrate, but these examples show the form of some children's talent.

"Emma loves all musical toys. She doesn't much care for TV, but when music comes on, she pays attention. She seems to really like the piano. I believe she will one day be a wonderful musician." 19 months, 2q37.1q37.3 deletion

"Millie loves music. She could whistle before she could talk." 4 years, 2q36.3 or q37.1 deletion

"Thom tunes into a track and can remember the tune for ages and recall the next time he catches it, he'll dance. He enjoys the Just Dance Wii game." 6 years, 2q37.3 deletion

"Brooke loves music and singing at school. Awaiting music therapy lessons." 6 years, 2q37.3 deletion

"Michaela knows the words to any pop song and sings very loudly." 11 years, 2q37 deletion

"She has a great singing voice and very good pitch. She plays the cello beautifully and composes her own songs on the piano." 12 years, 2q37 deletion

"He loves playing drums, and has an amazing gift for rhythm. He loves listening to music with drum beats, and goes to band class with his typical peers." 13 years, 2q37.1 deletion

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Support and Information

Rare Chromosome Disorder Support Group,
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Tel: +44(0)1883 723356
info@rarechromo.org | www.rarechromo.org

Unique is a charity without government funding, existing entirely on donations and grants. If you are able to support our work in any way, however small, please make a donation via our website at
www.rarechromo.org/donate
Please help us to help you!

This guide is dedicated to the memory of Monique A. Lock

On Facebook there is a group set up by a Unique member:
2q37 deletion - Rare Chromosome Disorder
There is also a Facebook community at 2q37DeletionSyndrome

Unique lists other organisations' 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 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. The guide was compiled by Unique and reviewed by Dr Rena E. Falk, clinical geneticist and cytogeneticist, Medical Genetics Institute and Department of Clinical Pathology and Laboratory Medicine, Cedars-Sinai Medical Center, Los Angeles, USA, and Professor of Pediatrics, Geffen School of Medicine at the University of Califonia, Los Angeles.

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