Support and Information

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

This updated information guide is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. The guide was compiled by Unique and reviewed by Dr Micheala Aldred, PhD DipRCPath, Assistant Professor at the Cleveland Clinic and Case Western Reserve University, USA and by Professor Maj Hultén, BSc, PhD, MD, FRCPath, Professor of Reproductive Genetics, University of Warwick, 2011. (PM)
A life with a 2q37 deletion

“David has been a great joy in our lives and has contributed a great deal to us and many others. We hope other children with his condition grow and flourish as he has.”

Clockwise from top left: 3 months; 10 years in pool; 10 years with brother and sister; 12 years; 39 years with nephew; 40 years; 37 years with father; 39 years with nephew.

References
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A life with a 2q37 deletion
“ Iain’s top qualification: a Gold Star from Daddy! ”

Clockwise from top left: first day home, about 5 weeks old; a few months old; probably around one year old; 2 years old; 14 years old; with father at 20 years old; 20 years old; 24 years old.

Sources and references
The information in this guide is drawn from what is known about 39 adolescents and adults with a 2q37 deletion. Some 23 adults have been described 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 at www.ncbi.nlm.nih.gov/pubmed. The full literature list is on page 15. The leaflet also draws on the experience of Unique’s members. When this leaflet was written, Unique had 16 members aged 15 or over with a 2q37 deletion, of whom 11 have a completed survey of the effects.


If you wish, you can obtain articles from Unique.

2q37 deletions in adolescents and adults
Chromosomes are structures inside the cells of the body that carry genetic information that tells the body how to develop, grow and function. They come in pairs, one from each parent, and are numbered 1 to 22 from the largest to smallest. In addition, there are two sex chromosomes (XY or XX). A 2q37 deletion is a chromosome disorder caused by a small bit missing from very near the end of one of the chromosome 2s. The other chromosome 2 is intact.

Each chromosome has a short arm (on the left in the diagram on page 4) called p from petit, the French word for small, and a long arm called q (on the right). A 2q37 deletion means that the chromosome has broken and a small bit is missing from very near the end of the long arm.

The karyotype or microarray report
The end of the long arm of chromosome 2 is divided into three bands: 2q37.1, 2q37.2 and 2q37.3. The missing section can include any or all of these bands. Generally speaking, people with a larger deletion do not do as well as people with a smaller deletion, but this is by no means a hard-and-fast rule.

People whose chromosome abnormality was detected by chromosome analysis, where chromosomes are directly visualised under a highly-powered microscope, are usually given a karyotype. Your geneticist or genetic counsellor will be able to tell you about the points where the chromosome has broken.

A karyotype for a 2q37 deletion could look something like one of these:

\[
46,XX,del(2)(q37.2)\\n
46: The number of chromosomes in your child’s cells
XX: The two sex chromosomes: XY for males; XX for females
del: A deletion, or material is missing
(2): The deletion is from chromosome 2
(q37.2): The chromosome has broken within the q37.2 band. The material from this point to the end of the chromosome is missing.
\]

As above but:

(q37.1q37.3) The chromosome has broken in two places: within the q37.1 band and again within the 2q37.3 band. The material between these two breakpoints is missing.

In the past very small deletions could be missed when looking down a microscope but today a technique known as array CGH or microarrays can identify tiny missing or extra pieces of chromosome material. Array CGH gives a very exact picture of what is missing and even a list of the known missing genes.
The results of a molecular analysis such as microarrays are likely to read something like this:

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arr cgh 2q37.3(238,922,192-242,391,504)x1
arr cgh
2q37.3

The specific DNA base pairs that are missing. Base pairs are the chemicals in DNA that form the ends of the ‘rungs’ of its ladder-like structure (see diagram, left). The first base pair shown to be missing is number 238,922,192 counting from the left of the chromosome. The last base pair shown to be missing is 242,391,504. Take the first number from the second number to get the number of base pairs missing. In this case, it is 3,469,312 base pairs. That can also be written as 3.5Mb.
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How old are the oldest people with a 2q37 deletion?
The oldest people in the medical literature are a brother and sister born in 1937 and 1944, seen when they were 61 and 54. The brother worked for several years in a sheltered workshop and was living with his sister in a residential home (Syrrou 2002).
The oldest member of Unique was born in 1968 and was 42 years old when this guide was written. He used to work for his local council on a special scheme and now works as a volunteer in two charity shops (Unique).

How can I find out how my child will fare in adulthood?
You can:

Read this guide. It tells you very generally how a group of adolescents and adults with a 2q37 deletion are getting on. But there can be big differences between your child and others in the group.

Compare your child with others with ‘the same chromosomes’. It may seem obvious to compare your child with others who apparently have the same amount of chromosome material missing. But the exact points where the chromosome has broken may be different, so they may have more genes missing or fewer. Even when people have apparently exactly the same or almost the same break points, individuals vary really a surprising amount.

Look at your child as an individual. By the time you are wondering how your child will get on as an adult, you probably already know quite a lot about them: any major health problems; any particular problems with development; the degree of learning support they need; and any difficulties they may have had with behaviour. These factors are very helpful pointers to their future.

Eyesight
Structural eye problems are rare but short and long sight, astigmatism and squint are all more common in people with chromosome disorders. Unique records suggest that short sight is especially common but is corrected with prescription glasses. Keratoconus has been reported twice. This is a degenerative condition where the cornea thins and is pushed outwards by the internal pressure of the eye. It is associated with chromosome conditions, occurs in both eyes, appears in the teens and needs to be monitored regularly (Wilson 1995; Felder 2009; Unique).

She doesn’t like bright sun; likes to wear sunglasses – 15 years

Hearing
Repeated ear infections are common in children with chromosome disorders and many are fitted with grommets [hearing tubes] to relieve glue ear, which causes a temporary hearing impairment. Some people with 2q37 deletions also have narrow ear canals but permanent hearing loss has not been recorded in the medical literature or seen in Unique.

Overreaction to common noises – 24 years

Dental care
In common with other adolescents and adults with a chromosome disorder, Unique members have a higher rate of need for dental treatment than other people. Three have needed teeth removed due to overcrowding; three have needed sealing treatments and one had additional fluoride. One teenager gets regular gum disease despite having his teeth cleaned carefully (Unique).
Spinal curve [scoliosis]
Spinal curvature may be more common than in typically developing children, due more to low muscle tone and muscle imbalance than to any vertebral abnormalities. It may need monitoring, bracing or surgery. By adolescence or adulthood, 4/12 Unique members have a spinal curve but none has required more than monitoring or physiotherapy and no adults have seen a deterioration (Unique).

Congenital hip dislocation
Congenital hip dislocation is a bit more common in 2q37 babies; it affected 2/12 Unique members. As adults, one walks with an unusual gait while in the other, whose dislocation was only found at the age of 13 months and who has also had dislocated kneecaps, it has had no effect on walking style (Unique).

Eczema
Eczema is common in people with a 2q37 deletion – but how troublesome is it? It affects seven/12 Unique adults and adolescents and can occasionally be severe and hard to control even with steroid creams. But the good news is that it seems to be better controlled with age. Some adults get it much less often than when they were children; others find it easier to control with standard creams and emollients or with diet; in others the eczema has resisted treatment but is ‘not serious’.

44 Slow healing eczema on my scalp which has left hair loss at the back of my head – 42 years

Asthma and other allergies
It is unclear whether children with a 2q37 deletion are more prone to asthma and other allergies than other children (Falk 2007). One of the two Unique adults who had asthma as a child still has it as an adult but it is well controlled with inhalers; the other has outgrown asthma. One teenager has hayfever and another has food allergies, requiring her to carry an Epipen [an adrenaline auto-injector] (Unique).

Other conditions
Adult and adolescent Unique members are by and large healthy, taking no regular medications and not needing regular hospital follow-up. Even a young man who had multiple heart, joint and gastrointestinal problems as a baby and child is a healthy young adult.

Anaemia has been seen in two adult men [developing at 38 years in one] and a teenage girl, but in all cases it responded well to iron supplements (Felder 2009; Unique). One man developed varicose veins at 30. A 16-year-old was diagnosed with Gilbert’s syndrome, a condition which can cause mild jaundice from time to time. It is usually harmless and does not require treatment and is due to a reduced amount of a chemical in the liver which processes a breakdown product of blood cells called bilirubin.

The exception to this general picture is a young woman who at 15 developed a laryngeal web which causes constant pain and stops her from eating, drinking and talking; at 18, she is fed by gastrostomy. She also developed gastro-oesophageal reflux at 15, which has since worsened and caused a hernia. She needs to sleep in a raised bed and in addition to anti-reflux medications, needs morphine for pain control (Unique).

Learning difficulties or disabilities: eventual outcomes
From what is known about development and learning, learning disabilities are variable but typically mild or moderate. Generally speaking, people with a slighter degree of difficulty have been able to go on to work either in paid or voluntary jobs, while adults with more marked learning impairments have needed all-round support as adults.

Opportunity is important: one Unique member who was in the ‘severe/profound’ class at a special school, though against expectation learned to read and write quite well, has held down a weekly voluntary job setting cutlery in a local restaurant.

More typically, a young man with mild learning difficulties who stayed in mainstream school for almost all his education and in addition to reading and writing well has a number of vocational qualifications [in cooking, woodwork, retail training, basic computing], now works as a volunteer in charity shops. Unique’s members with a more marked degree of disability have generally attended life skills courses at college after leaving school and then lived in residential homes or with their families. No Unique families reported any loss of intellectual ability with age.

44 She is expected to take at least a couple of GCSEs – 15 years, mild difficulties

Among particular skills and talents, a 42-year-old with mild difficulties lists getting on with people, compassion, caring, being thorough and good communication skills; a 20-year-old is good with computers; a 19-year-old with moderate learning difficulties has a marked degree of disability have generally attended life skills courses at college after leaving school and then lived in residential homes or with their families. No Unique families reported any loss of intellectual ability with age.

Artwork at a Unique meeting for 2q37 families - 15 years old
Independent living?
One Unique member lives independently in his own flat and shops alone on a daily basis but gets help with his weekly shop. The others either live at home or in residential care. Their personal care needs are very variable: most are independent in toileting, but one still needs pads. Families of adults say that their personal care skills – feeding, dressing, toileting, general hygiene – have stayed much the same since adolescence.

44 Since moving into a residential placement with seven other young adults, her communication and social skills have rocketed and she has started to speak with her family on the telephone – 19 years

Speaking and communicating
Most people with a 2q37 deletion but not all experience delay in learning to speak and to understand language. By the time they reach adolescence or adulthood, there is a broad range of communication ability. Some adults talk fluently with no perceptible difference between their understanding and their ability to express their thoughts. Others use signing, gestures, vocal sounds, facial expression and communication aids to express themselves. In line with a tendency toward autistic features, some use repetitive phrases and return repeatedly to the same topics. One 18-year-old stopped talking due to development of a laryngeal web. [The vocal folds in the voice box are usually separate, joining at a point at the epiglottis. A laryngeal web connects them with tissue that may be thin and translucent or thicker and fibrotic. Small webs rarely need treatment but larger ones can be removed surgically or by endoscopic laser.]

44 She has no formal speech but makes lots of her own sounds and has recently started to sing tunes and music such as Land of Hope and Glory and Chariots of Fire. She uses a few Makaton signs and is very good at communicating her needs, though people need to get to know her to understand what she wants. Her understanding continues to improve; she understands more than she can express – 25 years

44 He is unable to speak in an understandable manner, though communication may have slightly improved as an adult. He understands a lot more than he can say – 24 years

44 At 3 years she had no speech but was able to hum a tune without the words By 15, she was very vocal and giggling and shouting a lot, putting more sentences together and had good understanding. She did not look at you when she was talking and had facial contortions as if she was giving the matter careful thought. By 19 years, picture exchange was crucial in alleviating her anxieties and helping her cope better. She couldn’t pronounce words easily; and she used stock answers she had heard others say and remembered; sometimes she repeated what she had heard. Now she speaks fluently in full sentences but she still understands more than she can say – 19 years

44 Since she stopped talking eight months ago, she has used Makaton signing or she writes letters. If I don’t understand what she wants, she gets angry and stamps her feet. She understands most things and nods yes or no – 18 years

44 She can find it hard to express what she wants, especially when agitated. If speech is kept to short, precise questions her understanding and speech are the same; if too much information is given, her understanding goes – 15 years

which might be expected to reduce fertility. However, there have been no detailed studies of fertility in males with 2q37 deletions (Conrad 1995; Wilson 1995; Viot-Szoboszlai 1998; Reddy 1999; Syrrou 2002; Unique).

If my child has children will s/he pass on the 2q37 deletion?
Anyone with a 2q37 deletion who has children has a 1 in 2 (50 per cent) chance in each pregnancy of passing the condition on. To date there has been one unpublished report of a woman with a 2q37 deletion having a child, and the child was also affected. There have been no reports of men with 2q37 fathering a child. When the deletion is passed on, we do not know yet whether the clinical picture is similar in both parent and child.

Some special conditions

Those who have had heart surgery
About one person in five with a 2q37 deletion is born with a heart defect, usually a hole between the left and right sides of the heart. Three out of 12 Unique adults were born this type of heart defect; in one child the hole healed naturally, but two children needed open heart surgery. At 24 and 15 years old, all are healthy (Unique). Among 39 adults described in the medical literature, just one had surgery as a baby not only to correct a hole in the heart but also to correct a narrowing of the main blood vessel leading from the heart to the rest of the body [aortic coarctation], which can also be typical for 2q37 deletion. At 19, she was healthy (Fernández-Rebollo 2009).

Those who have had seizures
Between one in three to one in four children with a 2q37 deletion has epilepsy [seizures]. In most children there is no obvious underlying brain malformation associated with the seizures and they respond well to standard antiepileptic drugs (Falk 2007). Among 12 Unique adults, two had seizures as children. The seizures were well-controlled with medication in both: in one they resolved at the age of 10; in the other they were less troublesome by the age of 15. One of the two children was using words by 15, but not full sentences; both had at least a moderate degree of learning difficulty.

Kidney cysts
Children with 2q37 deletions may be more likely to develop kidney cysts. This is based on two children who had normal kidney scans when under two but were found to have kidney cysts later. No kidney cysts have been seen among Unique members. 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 (Conrad 1995; Falk 2007).

Wilms’ tumour
There are three reports of children with Wilms’ tumour who all had breakpoints at 2q37.1. Wilms’ tumour is a form of kidney cancer that affects children mostly under five. Most Wilms’ tumours respond well to treatment and most children can be cured. The most recent reviews recommend screening for children with breakpoints involving 2q37.1 or deletions encompassing 2q37.1. Wilms’ tumour is rare in adolescents and adults, so screening can be discontinued (Aldred unpublished).
earlier] in both boys and girls and there is a tendency for girls to develop a ‘pear-shaped’ build. Diet and exercise are usually enough to keep weight gain under control, but one mother reported that her daughter’s weight ‘ballooned’ nonetheless and low muscle tone makes it hard for adolescents to exercise with pleasure (Unique).

Most adolescents and adults eat a full range of foods but some still need their food cutting up small. A girl with a laryngeal web is fed by gastrostomy direct to the stomach but also takes soft food by mouth (Unique).

### Behaviour, mood, autism

Many children with a 2q37 deletion have no behaviour difficulties at all. Nonetheless, reports suggest that as children, around one in three people with a 2q37 deletion has an autistic spectrum disorder and autism has been diagnosed as late as 24 years. There are also individual reports of hyperactivity with attention deficit, intermittent aggression, obsessive compulsive disorders and sleep disorders. What happens as children mature?

As with children, a number of adolescents and adults are described with no behaviour problems at all. On the contrary, they are ‘gregarious and friendly’; ‘friendly and jovial’; ‘very friendly’; or have no behaviour problems (Chassaing 2004; Shrimpton 2004; Chaabouni 2006; Kitsiou-Tzeli 2007; Williams 2010). Other adults do have mood or behaviour difficulties, including depression; self harm; repetitive behaviours and aggression. These occur regardless of the size of the 2q37 deletion (Bijlsma 1999; Chaabouni 2006; Felder 2009; Williams 2010).

But reports in the medical literature are not very helpful as they rarely chart progress. What can Unique tell us? As reported in the medical literature, there is a group of adolescents and adults with no behaviour problems or no more than occasional outbursts and another group with behaviour difficulties acute enough to need professional help. Within Unique the group with no major behaviour difficulties is larger [5:3]. When difficulties do occur, they include outbursts of self harm; acute moodiness and withdrawal; attention-seeking; obsessive behaviours; aggression and worry about changes in routine. Two adolescents had behaviour problems that were so severe that the family could not cope and both are in residential care; the behaviour difficulties [tantrums, aggression, spitefulness, destructiveness] were obvious in both children from very early childhood. Behaviour problems are of course a common part of normal adolescence and there are at least three reports of behaviour improving with age and maturity. All the same, one adolescent experienced an unexplained behaviour deterioration, with more moodiness and withdrawal and another adult became more demanding with worsening self harm. Half of the Unique group have a diagnosis of an autistic spectrum disorder but some families comment that this exists alongside sociability.

Behaviour strategies that families have found useful include time out; avoiding caffeinated drinks; listening carefully to what their child wants; sticking to routines; and anger management. They have had behavioural support and training and two have had psychiatric referral and treatment with medication [risperidone].

### How about puberty?

The medical literature suggests that girls with 2q37 deletions start their periods at a similar age to unaffected girls, but both early and late puberty have occurred, periods are not always regular and may stop altogether. Among boys there is likely to be a similar pattern; one boy of 17 with late puberty has been seen (Wilson 1995; Power 1997; Shrimpton 2004; Chaabouni 2006; Chassaing 2004; Kitsiou-Tzeli 2007; Fernández-Rebollo 2009).

There is a suggestion from Unique that early puberty or early adrenarche (the early appearance of pubic and armpit hair and body odour without other signs of puberty) may occur, and family reports of Unique members show that this has occurred in two out of seven girls. Seven out of eight girls or women have disruptive premenstrual symptoms, regardless of age and whether or not they take a contraceptive pill. One young adult with hard-to-handle behaviour becomes ‘sad and aggressive’ around her periods. If this affects your daughter and she is taking a contraceptive pill, it may be helpful to discuss with her doctor using a different type (Unique).

### Do people with 2q37 deletions have fertility problems?

Girls with 2q37 deletions who go into puberty and have periods normally are likely to be fertile. In the medical literature, there have been reports of abnormal womb shape (bifid uterus) and abnormalities of the ovaries [dysgenetic] in women, which might be expected to reduce fertility. In men, there have been several reports of physical differences such as hypospadias [the hole normally at the end of the penis is on the underside instead], undescended testes, and small or abnormal [dysgenetic] testes.
Happy but an extremely short attention span – 15 years
She doesn’t have very good concentration skills and needs motivators to be changed regularly. She likes her own agenda and dislikes conforming. Aggressive behaviour is most obvious at school; she hurts others rather than herself. Destructive and unpredictable; she can’t be left alone at any time at home or school – 15 years
She can have a bad temper which she finds difficult to control. On one occasion she picked at her arm to make it bleed when she was very annoyed about something – 15 years

Sociability
The evidence from Unique is that people with a 2q37 deletion are sociable and friendly. They generally get on well with their family and others, although only one has a best friend and another, a young woman of 19, a boy friend. However, as you can see below, this is not always the case.
I feel that she is lonely. She lives in her own little world. She does join in games and activities but since she has stopped talking she can’t tell me how she feels. She doesn’t have friends and doesn’t seem a very happy little girl any more – 18 years

Sleep disturbances
One in three of Unique’s adult and adolescent members has a sleep problem but the problems vary in type, age of onset and progression. Three adults have always been poor sleepers; one finds it hard to get to sleep; another is awake for long periods at night. Two have been treated with risperidone, which helped one for a short time when she was younger but not as an adult. One child slept well until the age of seven when he became regularly aggressive and angry at bedtime. By the age of 13 he was better able to occupy himself and had become much calmer.

Mobility
How do the short, broad feet of many people with a 2q37 deletion impact on their function? How mobile are they as adults? All adolescents and adults known to Unique can walk and the great majority can walk as far as they want, even one who had surgery to re-align his feet as a baby and one who has one longer leg. Some walk slowly, some have a problem with depth perception, most need to be accompanied and occasionally supported. There is a tendency to tire easily, so a few sometimes use a wheelchair outdoors. None of the adults drives and only one uses public transport alone. Low muscle tone persists in some adults [and may underlie the rapid tiring] and they may have flat feet or feet that turn inwards. Mobility has lessened in adulthood for the oldest, who at 42 has fallen a few times and now uses a stick. All the same, he travels on public transport on his own, walks for pleasure, swims, goes to the gym and enjoys 10-pin bowling. Others enjoy riding, dancing, trampolining and cycling, although dancing can challenge coordination. An 18-year-old with a marked change in mood and behaviour since the age of 15 is also less mobile than she was and a 15-year-old has become less mobile due to a weight problem. Another 15-year-old complains of aches, especially backache, as well as tiredness when she walks a long way.

Most adolescents and adults point out that they need very wide shoes to fit their broad feet, sometimes with a very high arch, and that they have odd length toes, some of which may overlap. Some have specially fitted orthopaedic footwear and some switch between these and trainers or other shoes (Unique).

In addition to unusual toes, claw foot [pes cavus] alongside other deformities has also been reported, in this case leading to an unusual wide-based walking style. Two adults, both living in a home, were reported to have weak legs (Reddy 1999; Syrrou 2002).

Will my child be short as an adult?
Adult stature below the normal range seems to be more common among people with a 2q37 deletion but it is certainly not inevitable. There is a bell-shaped distribution of height in the general population. In people with 2q37 deletions, you get a similar distribution but shifted down a little, so some people are below what’s considered to be the normal range, others fall within the normal range and fewer come at the top end of the normal range. The medical literature suggests that children have a smaller growth spurt during puberty and stop growing earlier, so may be shorter as adults than expected from their earlier childhood growth.

Among eight Unique members who stopped growing between 13 and 18 years, three were the same height as the rest of their family, while five were shorter than other family members, two of them - one man, one woman - by a foot [30cm] or more. The others were 2-3 inches [5-7.5cm] shorter than other family members (Unique).

The published literature does not yet give final height data of adults and doesn’t compare them with their family, although one report does remark that two sisters were ‘short for their genetic height’. Three men were 59-68 inches [150-173cm] tall, while four women were 61-64 inches [155-162cm] tall (Wilson 1995; Syrrou 2002; Shrimpton 2004; Chaabouni 2006; Fernández-Rebollo 2009).

What about weight gain?
Overweight tends to increase with age among people with 2q37 deletions. The evidence from the medical literature is patchy: a 2-year-old girl was referred for a diagnosis to genetics because of obesity; a 17-year-old boy had ‘abdominal obesity’; some adults were markedly overweight for their height and some were in the top 10 per cent of the population for weight despite being relatively short. There is even less information about when weight gain tends to start, but in one girl the weight: height difference accelerated from puberty so that as an adult she was in the bottom three per cent of the population for height but the top three per cent for weight and her legs were particularly fat (Wilson 1995; Power 1997; Bijlsma 1999; Smith 2001; Syrrou 2002; Chassaing 2004; Shrimpton 2004; Fernández-Rebollo 2009; Williams 2010).

Evidence from Unique shows that a weight problem is common but not inevitable. Five/10 adult or adolescent members have no weight problem and some adults have a slight body build. Any weight problem tends to gather pace around puberty [sometimes