

7q32q34 deletions



Sources and references

The information in this guide is drawn from what has been published in the medical literature about babies, children and adults with a pure deletion from 7q32q34 without involvement of other chromosomes. 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 articles from *Unique*. The guide also draws on information on the Decipher database (<https://decipher.sanger.ac.uk>), on four unpublished cases from the Chromosome 7 database housed at the Hospital for Sick Children in Toronto (www.chr7.org), and on *Unique*'s database. When the guide was published, *Unique* had nine members with a 7q32q34 deletion (Brøndum Nielsen 1979; Stallard 1981; Sarda 1988; Verma 1992; Malmgren 2005; Judkins 2010; Chromo 7; Decipher; *Unique*).

Deletions on chromosome 7 between 7q32 and 7q34

People with a 7q deletion have some DNA missing from one of their chromosome 7s. The missing piece raises the risk of development and learning problems and physical abnormalities. But there is wide individual variation of what the impact may be.

Genes and chromosomes

Our bodies are made up of billions of cells, each containing about 30,000 genes. Genes act like a set of instructions, directing our growth and development and how our bodies work. Genes are carried on structures called chromosomes. There are usually 46 chromosomes - 23 inherited from our mother and 23 inherited from our father - so we have two sets of 23 chromosomes in 'pairs'. Apart from two sex chromosomes (two Xs for a girl and an X and a Y for a boy) the chromosomes are numbered 1 to 22, generally from largest to smallest. Each chromosome has a short arm (on the top in the diagram on page 3) called p from petit, the French word for small, and a long arm called q (on the bottom). For healthy development, chromosomes should contain just the right amount of material – not too much and not too little. People with a 7q deletion have one intact chromosome 7, but the other is missing a piece, which can affect development. Most of the clinical difficulties are probably caused by the presence of only one copy (instead of the usual two) of a number of genes. As a medium-sized chromosome, the 1150 genes on chromosome 7 represent about four per cent of the total number of genes in the human genome - the genome being the complete set of chromosomes in each cell. However, a child's other genes, environment (including things like diet, exercise, exposures, upbringing), and personal characteristics also help to determine future development, needs and achievements. Ultimately, the clinical outcomes will most likely depend on which of the genes in the 7q32q34 region are missing and which ones remain as duplicates.

How did the chromosome alteration happen?

When a sperm cell from the father and egg cell from the mother first join, each typically carries just one copy of each chromosome. Together they form a single cell that now carries two copies of each chromosome. This cell must make

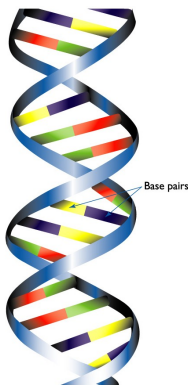
many copies of itself (and all the chromosomes and genetic material) in order to make the trillions of cells that form into a human during development. Sometimes during the formation of the egg or sperm cells, or during this complicated copying and replication process, parts of the chromosomes can break off or become arranged differently than usual. People with a 7q deletion have one intact chromosome 7, but a piece from the long arm of the other chromosomal copy is missing. It is believed that most of the clinical difficulties faced by someone with a 7q32q34 deletion are probably caused by having only one copy (instead of the usual two) of a number of genes. We are still learning the details about the specific jobs or functions of the genes that are sensitive to copy number (ie. 0, 1 or 2 copies). Also, it is important to stress again that a child's other genes, environment and unique personality help to determine their future development, needs and achievements.

Looking at chromosome 7q

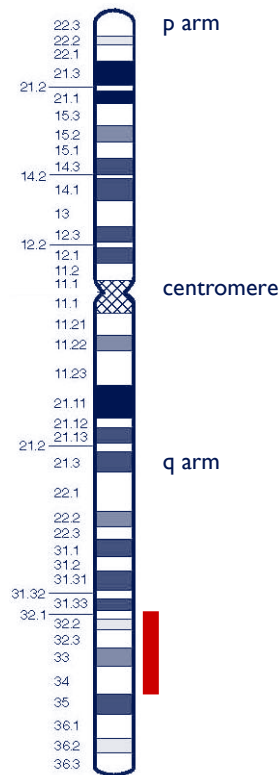
Chromosomes can't be seen with the naked eye, but if they are stained and magnified under a microscope, each one has a distinctive pattern of light and dark bands. Looking at chromosomes in this way, it is possible to see the points where the chromosome has broken and what material is missing, if the missing piece is large enough. A missing piece visible under the microscope is called a deletion.

In the diagram on the right you can see the chromosome bands are numbered outwards from the point where the long arm meets the short arm. Two-thirds of the way down the long arm, you can see five bands marked with a thick red line. These are bands 7q32q34. Band 7q32 is split into three bands (q32.1,q32.2,q32.3), then 7q33 and 7q34. Your child has lost some material from one, more or all of these bands.

Sometimes a deletion is so small that it can only be identified using molecular or DNA technology - in particular a technique using microarrays (array CGH). This technique shows in great detail gains and losses of tiny amounts of DNA throughout the chromosomes and can also show whether particular genes are present or not. A deletion so small that it can only be identified in this way is called a microdeletion.



Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.



DNA has a ladder-like structure. The chemicals that form each end of the 'rungs' of this ladder are called bases and since each rung has two ends, bases always come in pairs, known as base pairs, or bp for short. Base pair numbers are very long, usually in millions, so they are often shortened. For example, 1,800,000 base pairs is usually written as 1.8Mb. Mb stands for megabase.

Results of the genetic test

Your geneticist or genetic counsellor will be able to tell you about the points where the chromosome has broken. The results are likely to read something like this:

46,XY,del(7)(q32.3q34) dn

| | |
|------------|--|
| 46 | The total number of chromosomes in your child's cells |
| XY | The two sex chromosomes: XY for males; XX for females |
| del | A deletion, or material is missing |
| (7) | The deletion is from chromosome 7 |
| (q32.3q34) | The chromosome has broken in two places: one in band 7q32.32 and the other in 7q34 |
| dn | Short for <i>de novo</i> - Latin for 'new'. The parents' chromosomes have been checked and no change was found involving this region of 7q. The deletion is then very unlikely to be inherited and has occurred for the first time in this family with this child. |

Are there people with a 7q32q34 deletion who are healthy, have no major medical problems or birth defects and have developed normally?

So far, everyone whose chromosomes have been checked has been affected. But the effects vary and some people are only mildly affected.

Key features

People with a 7q32q34 deletion share some common features.

The most common features are:

- Developmental delay
- Language delay
- Variable impact on learning ability - most typically moderate or mild difficulties
- Behaviour concerns
- Early feeding difficulties
- Frequent infections (in children)
- Short stature (in around half)
- Some common facial features

These features do not affect everyone, and in any individual they can be more or less obvious.

■ Developmental delay

A delay in development is typical, so babies are late to reach 'milestones' such as sitting up and walking, handling toys, and talking. Children's developmental profiles vary, with some children more advanced in speech and language, others in fine motor skills and others in sitting and walking. First signs of developmental delay can usually be seen within the first six months and are often one reason for seeking a genetic diagnosis. But one boy with a deletion of bands 7q31 and 7q32 only raised concern when his speech failed to develop (Sarda 1988).

Babies and toddlers are likely to be late to hold their head steady, sit, move around and

walk, and also to handle toys. Having said that, children continue to make progress and some have very largely or entirely overcome their initial difficulties, although this will not be possible for all. *Unique* records show that babies learned to roll over between three and six months, and to sit between seven and 13 months (although one baby first sat at 20 months). They became mobile between 12 months and 16 months, although one baby was not moving until 32 months. The earliest mover had an unusual commando crawl and went on to walk early at 16 months, with a pronounced 'staggering gait'. Other children walked between 18 months and just over four years, but one four-year-old was not yet walking without support.

Once on their feet, some children quickly make up for lost time, while others retain a 'babyish' style of walking, and two children attracted a diagnosis of gait apraxia, a neurological disorder characterized by an inability to execute or carry out skilled movements, despite having the desire and the physical ability to perform them. One adult developed progressive balance problems, but we know no further details (Malmgren 2005; Decipher).

Underlying mobility problems in a few children is hypotonia, a low muscle tone, making babies feel floppy to hold and making it harder for growing babies and children to master purposeful control of their bodies. The evidence we have is that any hypotonia is likely to resolve fairly early in childhood, but that meanwhile children will benefit from regular physiotherapy (Verma 1992; Decipher; *Unique*).

Fine motor skills – hand use and hand-eye coordination – are also affected, creating difficulties in play skills, personal care (see below) and, later, at school. At least two children have attracted a diagnosis of dyspraxia - an impairment or immaturity of the organisation of movement. Children benefit from play therapy and occupational therapy, and some overcome their fine motor delay by the age of eight (Decipher; Brøndum Nielsen 1979; Stallard 1981; Sarda 1988; Verma 1992; Malmgren 2005; Chromosome 7; *Unique*).

“ Oliver’s initial developmental checks showed a six to nine month generalised delay. Today he is walking and running, pedalling a trike and climbing up and down the stairs ”
- 4 years, 7q33q34 deletion

“ By the age of three, his physical development was normal. Today he most enjoys swimming ” - 8 years, 7q33q34 deletion

“ Jack loves to swim but only in calm waters ” - 19 years, 7q32q34 deletion

“ As an adult, Cheryl has no major problems with stairs, running, jumping, cycling and absolutely loves dancing. She is slightly stiff and not very athletic. She lacks strength in her hands and fingers and her coordination is poor when her hands are out of sight ” - 32 years, 7q32q34 deletion

■ Language delay

Babies and children communication skills are typically delayed, but the evidence from *Unique* is that the majority do eventually communicate mainly by speech. They will be slower to learn and need more repetition than other children, as well as support from a speech and language therapist.

Unique records show that babies are late to smile – around three months. They

progress late to babbling, at around six months, and are significantly late in saying their first words. While one baby produced words at 16 months and another one month after his first birthday, the rest did not talk until they were three to five years old. Two children were not talking yet at the age of four, and a boy with a deletion from 7q31.2 to 7q32.3 wasn't talking yet at seven, though his understanding was good. We have less information on later speech development, but one girl was starting to form sentences at the age of 12.

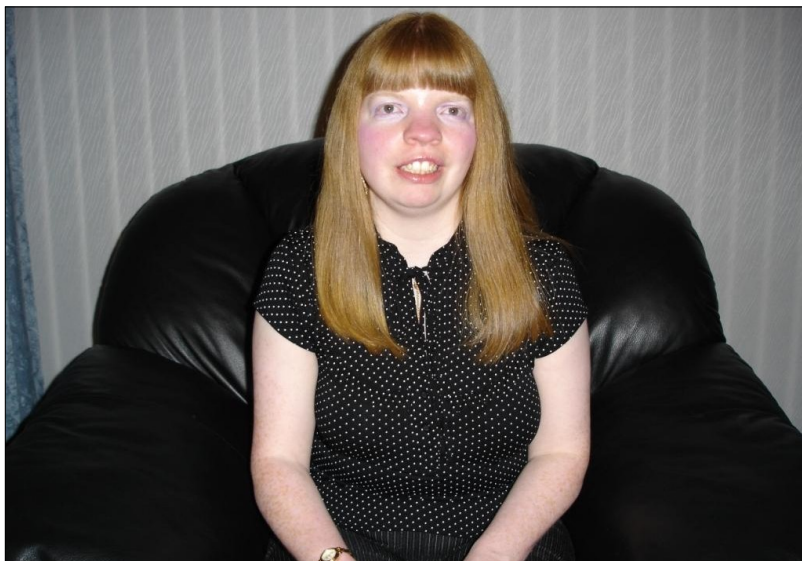
This evident delay means that children will be helped if they are encouraged to communicate in other ways. Signing helps to promote language acquisition and some children will do well with picture exchange systems or a communication device (Brøndum Nielsen 1979; Sarda 1988; Malmgren 2005; Decipher; *Unique*).

“ Oliver’s social interactions and non-verbal communication skills are advanced for his age ,while his speech remains delayed ” - 4 years, 7q33q34 deletion

His understanding is total but he cannot express himself verbally. He communicates by signing and in single words, and also has difficulty saying the letter *d* ” - 8 years, 7q33q34 deletion

“ Jack’s language is still quite difficult to understand but he persists and tries to explain in different ways ” - 19 years, 7q32q34 deletion

“ Cheryl can explain herself and get her point across, but she is probably not quite as fluent as others of her age. She has limited understanding so things must be explained thoroughly to her ” - 32 years, 7q32q34 deletion



■ Variable impact on learning ability - most typically moderate or mild difficulties

Children will benefit from early intervention and support with their learning throughout school. Children thrive on encouragement, praise and the desire to copy others, and

some have particular skills, such as a good memory. At the same time, children may have problems with paying attention and concentrating, and this can get in the way of their learning. Formal academic skills of reading and writing progress much further in some than in others, with a child with mild learning difficulties writing from the age of five while others hardly write at all. Most children reported in the medical literature or known to *Unique* have a moderate level of difficulty, but in a few it is mild and in one, severe. Most children attend a school for children with special needs, although children with mild difficulties may get enough support in a mainstream (regular) school environment. They will continue to need learning support as they move to college and, in at least some cases, on into the world of work. Outcomes can be very good, as you can read below (Brøndum Nielsen 1979; Malmgren 2005; Chromosome 7; *Unique*).

“ Jack works at the lower end of moderate learning difficulties. He has the cognitive ability of a five-year-old, and can read the odd word but has not started to write apart from his name ”

“ Cheryl gained six passes in our national exams as well as an office qualification. At school she was a very good reader with limited understanding. As an adult, she reads magazines and books and has an impressive knowledge of music, groups and singers and her information recall is good. Her writing is effective but the size of the characters can be inconsistent ” - 32 years, 7q32q34 deletion

■ Behaviour concerns

While some families voice concern over some aspects of their child's behaviour, this mingles with accounts of pleasing, loving behaviour and there is no evidence of a consistent pattern specific to a 7q32q34 deletion. The accounts below give a flavour of families' experiences.

“ Oliver is extremely outgoing, engaged and curious – a happy and fully involved little boy ” - 3 years, 7q33q34 deletion

“ Until around the age of three, he really only did what he wanted to do, but he then learned to do as he was told without tantrums. His behaviour has improved and is now normal for his age and he has no difficult behaviours. Socially, he would not play with other children until he was seven or so; today he still prefers to play on his own ” - 8 years, 7q33q34 deletion

“ Jack loves his mother very much and is always hugging her. He also loves his dog and gives it lots of cuddles. He is happy with his own company, and gets jealous if his siblings come back home to stay. He does not mix with other children because his social programmes have stopped with him being an adult. He often sits in cars and buses telling himself very imaginative stories out loud about where he comes from and about his real family who come from outer space. Recently he has become obsessive over the TV and computer and will often lash out if put under pressure to turn them off. He will still throw himself on the floor in a shopping mall when told he can't have something he wants. He has always tried escaping and running away, so it is difficult to contain him, but these days he does stay with his mother and is aware he could get lost ” - 19 years, 7q32q34 deletion

“ Cheryl was socially immature but being at work has improved her maturity and her perception of time. She enjoys music, reading, watching TV, and social outings; loves

music and dancing; and has an impressive CD collection. She has few friends but a good social life through her employers and likes animals and children. As an adult, she thinks of herself as happy, helpful, thoughtful, friendly, and caring. While she is generally good natured and sociable, she can also be too trusting and think of people she does not know very well as friends. At the age of 26, Cheryl was diagnosed with Obsessive Compulsive Disorder (OCD), which manifests itself mainly through the consumption of time. For example, it can take her the best part of an hour to get dressed. She will spend a long time hanging clothes up because they have to be perfect, which they will never be in her mind. She has had oral medication (antidepressants) from her psychiatrist and cognitive behavioral therapy (CBT) from her psychologist with regular visits from her psychiatric nurse, but with little success as the OCD is very extreme. Privately, she has also followed a neurolinguistic programme which with CBT had the desired effect, but Cheryl could not sustain the momentum for more than short periods ” - 32 years, 7q32q34 deletion

■ Early feeding difficulties

At least half of the babies known to *Unique* or reported in the medical literature faced early feeding difficulties, but while troublesome at the time, the difficulties were not severe enough in any baby to require a gastrostomy for direct feeding to the stomach, and all gradually improved. For some, sucking from the breast proved too great an effort and expressed breast milk was given by bottle, or babies switched to formula. Even sucking from a bottle was difficult for some, and in one case a squeezey bottle was prescribed. Others had a period of consuming less than they needed for healthy growth, known as ‘failure to thrive’, leading in one case to hospital admission and supplementation with energy-enriched formula.

A few babies had gastro-oesophageal reflux (bringing feeds back due to abnormal action of the valve between the food passage and the stomach). If careful positioning while feeding and lying with the head of the cot raised is not enough to control reflux, specially thickened anti-reflux milks can be prescribed for babies and young children and, where necessary, anti-reflux medication to help keep the stomach contents down and protect the inner walls of the food passage from stomach acid.

One baby got stomach ache on formula, so switched to cows’ milk at 22 months, while another had an allergy to cows’ milk. One baby declined all fluids by bottle, so was given them by syringe.

Feeding gradually improved in all babies, although one adult remains a ‘very picky’ eater, and another has difficulty swallowing tablets (Brøndum Nielsen 1979; Verma 1992; Decipher; *Unique*).

■ Frequent infections (in children)

Almost half the babies and children reported in the medical literature or known to *Unique* had frequent infections - usually coughs, colds and chest or ear infections. Some children suffered worse when ill than typically-developing children, and were more likely to end up in hospital. In two cases, the infections triggered seizures. There is no evidence of a lowered immune response in children with a 7q32q34 deletion and by the age of seven or eight, children had largely outgrown this tendency to frequent infections, so older children and adults enjoy generally good health.

■ Short stature (in around half)

There is a mixed and inconsistent picture of growth and eventual adult height in people with a 7q32q34 deletion. Although half the children and adults are shorter than expected for their family, this is much more evident in those reported in the medical literature than among *Unique* families. Adults known to *Unique* are average height (female) and 5' 7" (170cm, male), with normal build or very small. Reports in the medical literature show a mixed picture from the same family: while both adults reported are short, a 15-year-old boy from the same family is 5' 6" tall (167.5 cm). In another family, two six-year-olds are of normal height while a 13-year-old cousin is 12" (30cm) below the expected height for his age. A boy with a deletion between 7q31 and 7q32 is of normal height for his age: seven years. Two babies on the Decipher database were small and light at birth and continued to grow slowly; in the *Unique* cohort, this was seen only in a baby with a heart problem, and growth caught up after heart surgery (Brøndum Nielsen 1979; Sarda 1988; Malmgren 2005; Decipher; *Unique*).

■ Some common facial features

Children and adults with a 7q32q34 deletion may have certain facial features in common. In other ways, they are likely to look like other members of their family. *Unique* families are much less likely to remark on unusual facial features than researchers, who have identified common facial features - including widely spaced eyes, a long philtrum (the area between the nose and upper lip), unusually placed ears (most typically low set) and a large mouth with a thin upper lip. There is no consistent or characteristic look, and some people with a 7q32q34 deletion have other unusual facial features. These include a high or narrow forehead, prominent eyes, a flat or broad nasal bridge, a bulbous tip to the nose, a hooded eyelid (ptosis), tiny skinfolds across the inner corner of the eye (epicanthic folds), a downturned mouth, a small, short or upturned nose, a small or receding jaw and eyebrows that join in the middle (synophrys) (Brøndum Nielsen 1979; Stallard 1981; Sarda 1988; Malmgren 2005; Decipher; Chromosome 7; *Unique*).

Less common features

- Heart problems
- Epilepsy
- High palate or less often a cleft palate

■ Heart problems

There are no reports in the medical literature of heart problems associated with a 7q32q34 deletion. Four *Unique* babies were born with a heart problem, and a fifth developed a heart problem in adolescence, but the problems vary in severity and complexity. Two babies were born with a hole in the heart; in one case the hole sealed over by the age of six months; the other baby was monitored and neither baby has had a surgical repair. Another baby was born with a persistent ductus arteriosus that needed surgical correction. The ductus arteriosus is a channel between the aorta and the pulmonary artery that takes blood to the lungs and usually closes shortly after birth. When it stays open, the lungs receive more blood than they should and the heart has to work too hard. A fourth baby was born with Fallot's tetralogy: a complex heart

condition involving both a hole between the lower pumping chambers of the heart and an obstruction just below the valve in the artery that leads to the lungs. Blue (deoxygenated) blood cannot easily get to the lungs to pick up oxygen and some of it flows through the hole into the other pumping chamber, from where it is pumped around the body. Children with tetralogy of Fallot need a surgical repair.

A fifth *Unique* member developed a prolapsed valve in the heart in adolescence but is doing well without surgical repair.

■ Epilepsy

There are six reports of a child with a 7q32q34 deletion having a seizure or being diagnosed with epilepsy. In one 12-month-old baby the seizures were linked with a fever; and a six-month-old baby had two seizures, with no more by the age of eight. Two children are diagnosed with epilepsy - one developing seizures at the age of seven. Epilepsy in a *Unique* member is well controlled with anti-epileptic drugs (Brøndum Nielsen 1979; Judkins 2010; Decipher; *Unique*).

■ High palate or less often a cleft palate

There are five reports of babies or children with an unusually high palate or, in one case, a cleft palate that was surgically repaired. No *Unique* members have an unusual palate formation (Stallard 1981; Sarda 1988; Verma 1992; Decipher).



Other features

■ Spine

In three children, two of them adolescents, the spine developed a sideways curve (scoliosis). In three other children, the spine developed an inwards curve (lordosis). However, in the cases known to *Unique*, the curvature was not severe enough to need more than monitoring (Brøndum Nielsen 1979; Sarda 1988; Decipher; *Unique*).

■ Minor genital anomalies

Generally speaking, babies and especially boys with a chromosome disorder are more likely than typically-developing babies to be born with a minor genital anomaly. Out of 12 baby boys, two were born with undescended testicles. Treatment for undescended testicles depends on the suspected cause, but whatever it is, treatment is usually needed if the testicles do not descend naturally in time. The testicles can be brought down in a short operation under general anaesthetic called an orchidopexy. One boy was born with hypospadias, where the hole normally at the end of the penis is on the upper side instead. One was born with chordee - a downward curvature of the penis. If the anomaly is minor it can be left alone, but in a more severe case, surgical repair is called for (Malmgren 2005; Decipher; *Unique*).

■ Kidneys

There are four reports of children with a kidney problem. A six-year-old child had repeated urinary tract infections and was found to have kidney stones in the part of the kidney that collects urine and in the tube that leads to the bladder; the stones were removed. A newborn *Unique* baby had kidney reflux, where urine flushes back from the bladder towards the kidneys, but his kidneys were unharmed. A 19-year-old had ongoing kidney problems, requiring monitoring, but was well. A nine-year-old boy developed a kidney disorder known as CIq nephropathy, but it isn't known whether this is in any way linked to his 7q31.2q32 deletion (Brøndum Nielsen 1979; Judkins 2010; *Unique*).

■ Hernias

Two babies were born with an umbilical hernia. This shows as an abnormal bulge that can be seen or felt at the umbilicus (belly button). The hernia develops when a small opening in the abdominal muscles that allows the umbilical cord to pass through does not close after birth. Part of the lining of the abdomen, part of the intestine and sometimes fluid from the abdomen passes through the opening, causing the hernia. Many umbilical hernias close naturally by the age of three or four but a very large hernia or one that stays open after this age can be closed surgically. One of the babies had the hernia closed at 13 months.

Another baby was born with an inguinal hernia. This shows as a bulge in the area where the lower abdomen meets the upper thigh (the groin). The cause is that an opening in the lower part of the wall of the abdomen that is open during fetal life but closes before birth, does not in fact close. The remaining opening may be small, only allowing fluid through, or it may be large enough for something such as a loop of the intestine or another organ to get stuck in it. An inguinal hernia should always be assessed by a doctor and may need surgical repair (Brøndum Nielsen 1979; Stallard 1981; Malmgren 2005; Decipher).

■ Eyesight

While most children and adults with a 7q32q34 deletion have perfectly good eyesight, a few have unusually long or short sight, corrected with glasses, and a few have strabismus (a squint). In strabismus, the crossed eye can look inwards, outwards, up or down. The main effect is that usually the child will have one stronger eye because the brain gives priority to one eye, with the result that the weaker eye does not 'learn' to

see as well. Treatment can include patching the stronger eye, exercises, drops to encourage the use of the weaker eye, glasses to correct a refractive error such as long sight, and surgery to realign the muscles that hold the eye in place.

Two children in the medical literature have optic atrophy, where there is some loss of vision due to a proportion of the nerve fibres linked to the optic disc being lost, but this has not been seen in *Unique* (Brøndum Nielsen 1979; Malmgren 2005; Decipher; *Unique*).

■ Hearing

Most children and adults with a 7q32q34 deletion, including all those known to *Unique*, have perfectly normal hearing. There is just one report of a 15-year-old with some permanent hearing loss. However, the high rate of upper respiratory tract infections in babies and young children puts them at risk of developing the temporary hearing loss known as glue ear as a consequence of repeated ear infections. While glue ear will resolve naturally in time, many children will need a grommet (a small ventilation tube) inserted into the eardrum.

■ Personal care

Babies and toddlers are typically late to acquire the skills that underpin personal care. They are immature in using and coordinating their hands, and learn to grasp, let go and hold on to utensils later than typically-developing children. Toilet training is usually also delayed but evidence from *Unique* suggests that it is achieved between 22 months and five years. One four-year-old was not yet toilet trained. This delay means that children will need help with daily tasks of dressing, undressing and washing for longer than other typically-developing children (Sarda 1988; Verma 1992; *Unique*).

“Cheryl is still a little uncoordinated, so she needs help with personal care (bottle opening, hairwashing etc)” - 32 years, 7q32q34 deletion

■ Hands and feet

While it is relatively common for people with a chromosome disorder to have slightly unusual hands and/or feet, this is not very obvious in people with a 7q32q34 deletion. Out of seven people on whom we have information, two have entirely normal hands; three have small, short and sometimes broad hands; one has large and broad hands and one has a thumb with three joints, like a finger (Brøndum Nielsen 1979; Stallard 1981; Sarda 1988; Malmgren 2005; Chromosome 7; *Unique*).

Feet, too, are usually entirely normal, although one baby was born with a mild case of club foot (talipes equinovarus) that was treated successfully (Brøndum Nielsen 1979).

Pregnancy, birth and newborn

We have some information on pregnancy for 10 mothers and babies. Four pregnancies were entirely normal and the baby was born around the due date. Two mothers experienced some bleeding and one noticed that the baby moved very little. Two babies were marginally premature, born at 37 weeks, and one was overdue at 43 weeks. One was feet first in breech position and born by Caesarian section. In most cases, the baby's delivery went smoothly, but one mother had a difficult labour with a small placenta and very short umbilical cord.

Birthweights for babies born around term range from 2.6 kg (5lb 12oz)-3.59 kg (7lb 15oz). On the whole, babies were rather small for dates - on average 2.9 kg (6lb 6oz), or around 550 g (20 oz) lighter than an average term baby.

While one or two babies seemed entirely normal at birth, one failed to cry and another had episodes where he turned blue and his hands and feet twitched. The great majority for whom we have any information had difficulties feeding and putting on weight in the early months (see Feeding). (Brøndum Nielsen 1979; Stallard 1981; Sarda 1988; Malmgren 2005; Decipher; *Unique*).

Why did the chromosome deletion occur?

A blood test to check both parents' blood chromosomes is needed to find out why the 7q deletion occurred in the child. Most 7q interstitial deletions occur when both parents have normal chromosomes. The term that geneticists use for this is dn, short for 'de novo', meaning 'a new event'. Dn 7q deletions are caused by a sporadic mistake that is thought to occur when the parents' sperm or egg cells are formed or very soon after fertilisation [see page 3].

No environmental, workplace, dietary or lifestyle factors are known to have caused these chromosome changes. What is certain is that as a parent there is nothing you did to cause the break to occur and nothing you could have done would have prevented it from occurring in your baby. No one is to blame when this occurs, nobody is at fault and there is no reason for anyone to feel guilty.



Can it happen again?

The possibility of having another pregnancy with a 7q deletion depends on the parents' chromosomes. When both parents have normal chromosomes, the deletion is very unlikely to happen again. If either parent has a chromosome change involving 7q, the possibility is greatly increased of having other affected pregnancies. If they wish, parents should have the opportunity to meet a genetic counsellor to discuss the specific recurrence risks and options for prenatal and preimplantation genetic diagnosis (PGD). PGD requires the use of in vitro fertilisation and embryo biopsy, and only healthy embryos are transferred to the mother's uterus. If the parents choose to conceive naturally, prenatal diagnosis options include chorionic villus sampling (CVS) or amniocentesis to test the baby's chromosomes. Testing is very accurate, although not all of these tests are available in all parts of the world.

Could my child with a 7q deletion have similarly affected children?

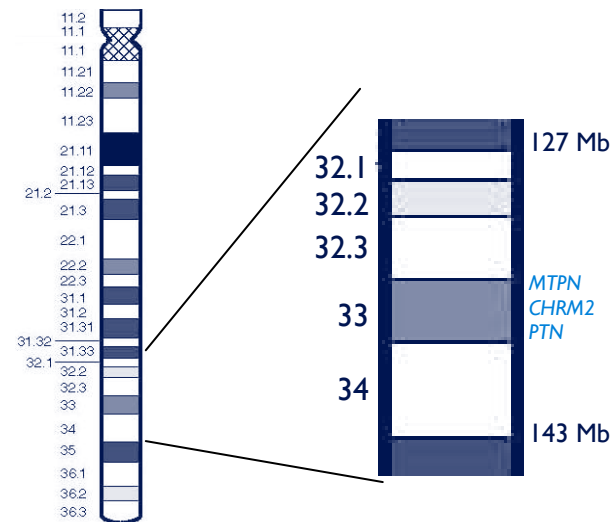
In any pregnancy, someone with a 7q deletion is likely to have a 50 per cent risk of passing it on and a 50 per cent chance of having a child without it. If the deletion is passed on to a second child, it is likely that he or she will have characteristics similar to the first child but there could also be significantly different clinical presentations for the reasons described earlier.

Some genes in the deleted area

MTPN Involved in the development of the nervous system: plays potential role in cerebellar morphogenesis and differentiation of cerebellar neurons (Malmgren 2005)

CHRM2 Involved in the development of the nervous system: mediates various cellular responses and has been implicated in neurogenesis (Malmgren 2005)

PTN Involved in the development of the nervous system: has neurite extension activity (Malmgren 2005)



Chromosome 7

Notes

Inform Network Support



Rare Chromosome Disorder Support Group,

G1, The Stables, Station Rd West, Oxted, Surrey. RH8 9EE

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Please help us to help you!

This leaflet 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. The information is believed to be the best available at the time of publication. It was compiled by *Unique* and reviewed by Professor Stephen Scherer, Director, Centre for Applied Genomics, Hospital for Sick Children, Toronto, Canada; and by Professor Maj Hultén, University of Warwick, UK 2012.

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