



Support and Information

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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 and has been verified by Dr Steve Scherer, The Hospital for Sick Children, Ontario, Canada and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK. 2005.

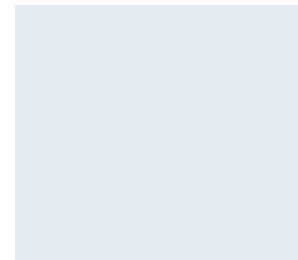
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Duplications of 7q



Sources & References

The information is drawn partly from the published medical literature. The first-named author and publication date are given to allow you to look for abstracts or original articles on the internet in PubMed. The leaflet also draws on *Unique's* confidential database. At the time of compiling the information, *Unique* had 29 members with a 7q duplication, nine of them with a pure duplication of 7q that did not involve any other chromosome arm. Eight families completed a questionnaire about their child's development in 2005. References to information from *Unique* are marked U. If you have a family member with a 7q duplication or are a health professional and would like to add your experience to the *Unique* database, please contact info@rarechromo.org

7q duplications

A 7q duplication is a rare genetic condition, in which there is an extra copy, known as a duplication, of part of the genetic material that makes up the body's chromosomes. Generally speaking, having extra chromosome material does make it more likely that a baby will have birth defects and experience growth and developmental delay but the outcome for each child can be quite individual. The precise effects of gaining material from a chromosome vary depending on how large the duplication is, how many genes the duplication contains and what those genes do.

Chromosomes are the microscopically small structures in the nucleus of the body's cells that carry genetic information. They are numbered in pairs from 1 to 22, running approximately from largest to smallest, with one member of each pair coming from the father and one from the mother, in addition to the sex chromosomes, X and Y for a boy and two Xs for a girl.

Each chromosome has a short (p) and a long (q) arm. People with a chromosome 7q duplication have a duplication of some of the material from the long arm of one of their chromosome 7s.

Main features

Only two babies have been described with a duplication (called a trisomy) of the entire long arm of chromosome 7.

In babies and children with a smaller 7q duplication, these are the most common features. Many of these features are also common in children with other chromosome disorders.

- Developmental delay
- Some degree of learning difficulty or disability
- Low muscle tone, so the body feels floppy
- Low birthweight and slow weight gain in babies. Eventual height is often short
- High palate (roof of the mouth). There may be a split (cleft)
- Large head and, at birth, a very large soft spot (fontanelle)
- Most babies are healthy at birth, although there may be a heart condition
- Unusual position or angle of one or both feet (talipes, club foot)
- Curvature of the spine
- Subtly unusual facial features

(Bartsch 1990; Ndah 2000; Back 2001; Lukusa 2002; Rodriguez 2002).

Can it happen again?

The chances of having another child affected by a 7q duplication depend on whether the duplication is inherited or not. If a chromosome analysis shows that either parent has a rearrangement of their own chromosomes, they have a significantly increased risk of having another affected child.

If the chromosomes of both parents are normal, it is very unlikely that it will happen again.

There is an extremely slight possibility that in some people the duplication occurred during the formation of the cells that later give rise to the egg or sperm. This can result in a mixture of normal egg or sperm cells and cells with unbalanced chromosomes (gonadal mosaicism or germline mosaicism). When this occurs, there is a tiny but real chance that parents with apparently normal chromosomes could have another affected pregnancy.

When you are ready to think about another pregnancy, you should be able to discuss your individual situation with your genetics service and weigh up the pros and cons of prenatal diagnosis.

Causes

Rearrangements in chromosomes occur as part of evolution. They affect people from all parts of the world and from all types of background. They also happen naturally in plants and animals. So there is no reason to suggest that your lifestyle or anything that you did caused the duplication of chromosome material.

To answer the question "Why did this happen?" a geneticist will first need to know about the parents' chromosomes. A blood test will give the answer and in around half the children with a 7q duplication, it will turn out that one parent, believed usually to be the mother, has a structural rearrangement of their own chromosomes.

When it's inherited

There are three types of typical chromosome rearrangement in the parent that are likely to give rise to a 7q duplication in a baby. In these rearrangements, the parents themselves are usually healthy and have no developmental problems because the correct amount of chromosome material is present.

A parent with a **balanced translocation** has chromosome material switched between different chromosomes. Usually two chromosomes are involved, but it can be more.

If the short arms of either chromosome 13, 14, 15, 21 or 22 are involved, the effects on the baby are most likely to just be of the 7q duplication. If the swap has taken place with the long arm of any of the chromosomes or with the short arm of any other chromosome, it is likely that there will be additional effects caused by material lost from that chromosome. Your geneticist can tell you about the detail of this.

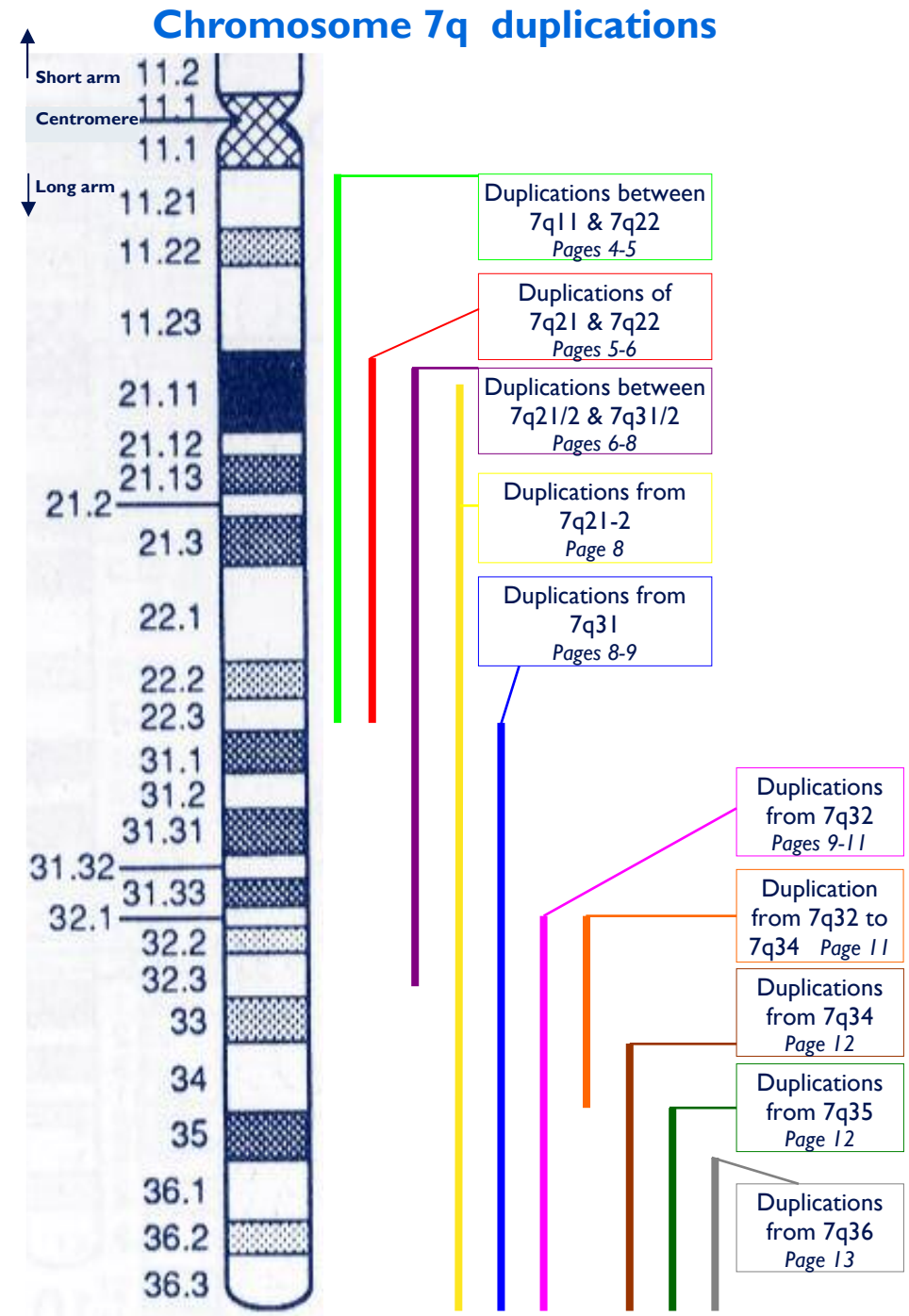
A parent with an **insertion** has a piece of 7q inserted into a different chromosome. As the segment of 7q is missing from one chromosome 7, the parent has the correct amount of chromosome material.

A parent with an **inversion** has one chromosome 7 in which two breaks have occurred. The broken length of the chromosome has flipped 180 degrees and reattached itself to the two broken ends. This too can lead to a 7q duplication in the baby.

In all inherited cases, the risk of having another affected pregnancy is significantly increased. Families can discuss their individual situation with their genetics service.

When it's not inherited

When the tests on the parents' chromosomes show they are completely normal, the duplication has most likely arisen as a chance event. Geneticists call this *de novo*, meaning that it is not inherited and the affected child is the first person in the family with the chromosome disorder.



Duplications between bands 7q11 and 7q22

Four babies or children have been described, each with a slightly different duplication: 7q11q22, 7q11.21q11.23, 7q11.22q11.23 and 7q11.23q21.2. The pattern of unusual features was different in each and incomplete descriptions mean that no features are known to be consistent for all, apart from a low normal birth weight (range 2500g to 3269g, 5lb 8oz to 7lb 3oz).

Taking the four youngsters as a group, features reported for two included a large head with a large soft spot (fontanelle) and wide open sutures (seams between the bone plates of the skull, on top of the head), but while in one baby the head was only slightly larger than normal and the fontanelle closed by the age of two years, another baby had marked hydrocephalus (excessive fluid within the brain).

Two babies were described as having a prominent forehead and in one the seam line between the skull plates running down the middle of the forehead was raised. Two babies had a cleft (split) in the roof of the mouth (palate) and one had a cleft lip; another had a high palate. Three babies had noticeable genital involvement: in one girl the vagina, ovaries and uterus failed to develop and in one boy, the testicles failed to descend and on one side was removed surgically. On this side the vas deferens that takes sperm from the testicle was unusually long and narrow. In a second boy, the testicles were not descended at birth and the penis was small. Two children experienced seizures and one had unexplained intermittent feverish episodes as a baby. Feeding difficulties and rather slow weight gain were described in three children, as was constipation.

The differences between these children were as remarkable as the similarities. While one child had healthy major organs (brain, heart, lungs, kidneys) but a loose hip, a missing rib and premature growth of pubic hair, another had marked hydrocephalus and a small amount of brain tissue, malposition of the heart and a failure of the left eye to develop. Another child had an undeveloped left kidney and a markedly sunken eye and developed aseptic necrosis of the right hip, a condition where a poor blood supply causes an area of the bone to die.

In terms of development, one child was sitting up at 10 months and walking by the age of three but had not yet started to speak at the age of four. Another child showed more marked delay in reaching his mobility milestones (Hoo 1982; Kardon 1983; U).

Zackary

Zackary's chromosome disorder was first suspected in early pregnancy when a nuchal translucency scan showed excess fluid around his neck and abdomen. The rest of the pregnancy was uneventful and he was born at 39 weeks, a healthy size and weight. Zackary's most obvious difficulty at birth was his cleft lip and palate but he had other more subtle signs – his main fontanelle was large and his forehead was prominent, his ears were set low and rotated backwards,

Duplications from 7q36 to the end of the chromosome

There is little experience with individuals with a small duplication of material near the end of the chromosome, but it appears that effects are less far-reaching than when the duplication is larger. Common features include:

- Some degree of developmental delay, especially speech
- Large head with a prominent forehead

(Lowry 1983; Verma 1992; U)

Oliver

As a new baby, Oliver gave no cause for concern: his Apgar scores were normal, he weighed a healthy 9lb 7oz/4280g when induced at 11 days after term and he latched on well to the breast and fed for a long time. The family doctor had a concern that Oliver's very large head might mean he had hydrocephalus, but both CAT and MRI scans showed ventricles in the brain that were enlarged but within normal limits.

The first signs that Oliver was more than a very large and beautiful baby lay in his behaviour and his development. He liked nothing more than to lie still on his playmat with his rattle. Oliver's muscle tone was low and this, together with tightly contracted joints in his ankles, feet and neck delayed his milestones. In his pushchair as a baby, he could not hold his position and would slide to one side. He learned to sit at 11 months; by 13 months he could shuffle and by 24 months he could walk without support. At the age of almost 10, he wore inserts in his footwear and support boots to prevent his ankles from rolling in, and because he could still trip and fall when he was tired and floppy, he wore a helmet for outdoor play. Oliver has been able to eat with a spoon since he was four, but has not yet mastered a knife and fork.

When Oliver wanted something, he would tap your arm and pull you. He used signs, vocal noises and single words, trying to sign at the same time. Yet he understood almost everything. In terms of learning, he needed considerable support and attended a special school where he learned best in a small group. Being outgoing, friendly and caring helped him to learn.

Oliver loved music – listening to it, watching it on TV and strumming his guitar. He loved the computer and being and playing with other children and he liked being outdoors and digging in the garden. His behaviour was only difficult when he was tired.

Oliver has a small duplication between 7q36.1 and 7q36.3.

Duplications from 7q34 to the end of the chromosome

Twelve children have been described in the medical literature, most of them also with a chromosome deletion from an unbalanced translocation. Only three have been described with a 'pure' 7q34 duplication (with no simultaneous loss of material from another chromosome). Of *Unique's* two members with a 7q34 duplication, both had a deletion but in one, the effects of duplication were more important. It appears that the most frequent features linked with the duplication, all of them common in children with other chromosome disorders, are:

- Developmental delay and low muscle tone (hypotonia)
- A degree of learning difficulty or disability
- A relatively large head with a large main fontanelle (soft spot)
- Birthweight within the normal range (6lb 8oz/2948g to 7lb 9oz/3422g at term)
- Feeding difficulties caused in part by low muscle tone and difficulty in sucking
- Spinal curvature, (kyphoscoliosis, so the spine curves forwards and sideways)
- In boys, minor genital anomalies including undescended testicles
- A recognisable pattern of facial features, including a short nose with a low bridge, an open mouth, a short neck and sometimes irregular teeth.

Individuals have been described with particular features, notably a type of glaucoma known as buphthalmos. In this condition the pressure inside the eye rises unusually high; treatment usually involves surgery to drain excess fluid. Other features include strabismus (squint) which can be treated with patching or surgery, webbed or unusually jointed fingers, toes and thumbs; dislocated shoulders and a missing set of ribs (Forabosco 1988; Romain 1990; Kato 2001; U).

Duplications from 7q35 to the end of the chromosome

Features appear to be quite variable. Most individuals described in the medical literature also have a deletion from another chromosome as part of an unbalanced translocation and the apparent variability may be due to this or to minute differences in duplication size. The most common features include these:

- Developmental delay
- Low muscle tone, sometimes co-existing with lower limb hypertonia (high muscle tone)
- A variable degree of learning difficulty
- Unusual genital features in boys, such as undescended testicles
- Normal birth weight and growth rate before and after birth
- Nystagmus (irregular eye movements)
- Foot anomalies
- Unusually long fingers
- A pattern of subtle but unusual facial features, including a relatively short neck, low set ears, a low nasal bridge, a small nose, a prominent forehead, a relatively large head and skin folds over the inner eye corner of the eyes (Hoo 1995; Speleman 2000; Lukusa 2002; Morava 2003).

his eyes had an upwards slant and were unusually wide apart. Investigations also revealed a heart murmur, which resolved naturally, and undescended testicles, which would be operated on surgically.

The cleft palate and lip made feeding very difficult at first and Zackary was fed with a squeezey bottle until his hard palate and lip were repaired at three months and his soft palate six months later. However, Zackary brought feeds back up his food pipe and a barium swallow test showed that he tended to aspirate them. To control this condition, known as reflux, he was fed sitting upright and his bottles were thickened to a custard consistency. All the same, at the age of 2, he still found it very hard to gain weight.

Zackary's first year was punctuated by ill health, with monthly hospital stays for respiratory infections, ear infections requiring grommets (tubes inserted into the eardrum to improve hearing) and urinary infections requiring protective low dose antibiotics. At the age of one, he had a febrile convulsion and at 15 months, a suspected breathing arrest.

Despite his multiple setbacks, Zackary has made steady progress. He could roll by the age of 1 and at 2 could take his own weight in a walker. His mobility was hampered by his naturally low muscle tone as well as his unusually flexible joints. He could sometimes tap a toy or his tray to show his needs and loved to tap on a keyboard. At the age of 2, Zackary was not yet very responsive to sound or to visual stimulation and his predicted need for support with his learning was considerable. Nonetheless, he had a happy, smiling, laughing nature and was a 'fabulous' sleeper.

Small duplications involving 7q21 and 7q22

Three individuals have been described, each with a slightly different duplication: 7q21.2q22.1; 7q21.2q22.3; and 7q21.3q22. Despite incomplete reports, the evidence is that these features - all of them frequently found in youngsters with a rare chromosome disorder - were common to all three:

- A degree of developmental delay. Two boys were walking by the age of 2
- Learning difficulties, typically moderate
- Initial feeding difficulties and failure to thrive

Features seen in two individuals included:

- Low muscle tone (floppiness) and unusually flexible joints
- Recurrent viral or bacterial respiratory infections in childhood, leading to frequent hospital stays

- Strabismus (squint), corrected by patching in one child
 - Short stature, with a relatively large head
 - Some behaviour difficulties and problems with social integration, formally diagnosed in one child as autism. One child had a tendency to be aggressive
- Observations in one of the three included: a cystic kidney at birth; missing teeth and poor enamel development; multiple tissue joins between the gums and cheeks; webbed fingers (Lukusa 1998; U).

Duplications between bands 7q21 or 7q22 & 7q31 or 7q32

Nine people have been described, mostly as babies, with duplications of varying sizes between 7q21 and 7q32. The main features are all common in children with other chromosome disorders.

- Variable degree of developmental delay
- Low muscle tone and unusually flexible joints
- Growth delay
- Typical facial features include a rounded forehead, small eye openings, widely spaced eyes with skinfolds across the inner corners, squint (strabismus) and low set ears.

At birth, most babies were an appropriate length and weight for their gestation. Typically, they found it difficult to put on weight and needed feeding support, in some cases including high calorie supplements and direct feeding into the stomach through a gastrostomy tube. Little information is available on adult build, but one teenager was both tall and well-built. Two *Unique* members showed signs of lactose intolerance and thrived on a lactose-free diet, although this may be unconnected with their chromosome disorder.

All children for whom information was given suffered frequent respiratory infections as babies. Ear infections were common as well and were associated with glue ear and a moderate hearing loss relieved by grommet (tube) insertion. Two children had narrow ear canals, in one case needing surgery. A further child had a malformed bone structure within the middle ear and permanent hearing loss on one side. In terms of vision, most children had a squint (strabismus), correctable typically by patching or surgery and one child was born with cataracts (see page 7). The vital organs – brain, heart, lungs – were usually healthy but one child was born with an absent left kidney and a malformed right kidney.

Developmentally, children showed a mild to moderate level of delay and were markedly affected by their low muscle tone (floppiness) and hypermobile joints. In one child the hip sockets were shallow and she wore a harness as a young baby to develop them.

In terms of learning, all children showed some level of delay but in some children at least, language was relatively well preserved compared with mathematical skills. One child developed the vocal tic disorder Gilles de la Tourette syndrome (Novales 1982; Romain 1990; Humphreys 1991; Kroisel 2001; U).

and chin, long toes and clenched fingers, she had few of the 'typical' facial signs of a 7q duplication. Her head was not large, her fontanelle was not wide open and she did not have a prominent forehead. She would be about five years old before the diagnosis was made that she had material from 7q, with a breakpoint at 7q32, attached to chromosome 5.

In terms of feeding, Kim was a slow starter. She had no sucking ability and was initially tube fed but progressed to a bottle by four weeks. At the age of 19, she was eating well although she had a continuing problem with constipation and remained very small for her age.

Healthwise, Kim's hips have caused the greatest problems, involving two surgical operations at the age of 7 and further surgery ten years later to remove plates and pins. She also had surgery at the age of five to lengthen her Achilles tendons. This naturally impacted on her mobility and while she had physiotherapy from the age of two, and did walk for a while using an orthotic walker, at 19 she was no longer able to take her own weight. To move around, Kim bottom-shuffled indoors and used a wheelchair outdoors.

During puberty, Kim had four seizures, but they have not needed control with medication. She has also had a dilated ureter and mild hydronephrosis (the kidney and the tube leading to the bladder were swollen) but apart from this, Kim has been healthy.

Developmentally, she has made progress, although as a young adult she still needed others to hold her drinking bottle and help her hand over hand to feed herself with a spoon. She used pictures, signing and vocal noises to communicate but was more strongly motivated to sit and watch the world go by. Kim has had difficult times when she has pulled her hair out and banged her head, apparently because it gave her pleasure and she wanted attention. She wore a protective helmet in bad spells.

Kim has a beautiful smile and an engaging personality and has clear likes and dislikes, her family says. She is cheeky, loves a cuddle, enjoys flirting and music of any kind. At 19, she left school and moved on to Day Services where she was very happy and enjoyed getting out and about.

Duplication from 7q32 to 7q34

A child who was born with a squint was found to be missing one of the six muscles that hold the eyeball in correct alignment. The lateral rectus muscle was missing on the right side and underdeveloped on the left side. One of the other muscles involved in holding the eye straight (the inferior oblique muscle) was also smaller than normal. In addition, the child had some degree of developmental delay and was short for his age. His optic nerves were underdeveloped, causing quite low vision (Keith 1988).

of puberty and irregular periods and once in a girl with heavy and regular periods. (Newton 1972; Bass 1973; Schmid 1979; Novales 1982; Couzin 1986; Talley 1989 Bartsch 1990; Zackowski 1990; U).

Marte

Within minutes of her birth in 2002, Marte developed breathing problems. She had other unusual signs – her head was large and the soft spot on the top was also wide open, she had small tags beside each ear and her fifth fingers, although apparently normal, were missing a joint. However, it was Marte's breathing problems that led to the discovery that she had two heart defects – coarctation of the aorta (narrowing of the vessel taking blood from the heart to the body) and a ventricular septal defect (a large hole between the lower heart chambers). Paediatric surgeons corrected the heart problems when Marte was just six days old. Chromosome tests showed that she had a duplication of 7q from band q32 and a deletion from band q35 of chromosome 5.

Healthwise, Marte has had some problems, and was in hospital fifteen times in her first three years with chest infections and pneumonia, as well as five times for an inflammation in her left thumb. Her large head size has been investigated and a diagnosis of communicating hydrocephalus (excessive fluid) has been made, although Marte needs no treatment for this. With daily salbutamol inhalations to improve her airways function, she is now in good health.

Developmentally, Marte has made steady progress, learning to roll at 8 months, sitting on her own at 17 months and bottom shuffling at the age of two. She has regular physiotherapy and uses both a stander and a walker, as well as a pushchair outdoors. In terms of hand use, she could use both her hands together at eight months, hold her bottle by the age of 19 months and by 2½ years old, was using a spoon, waving hello and goodbye and clapping.

By the age of three, Marte was alert and interested and could tell her family her age, using her fingers. To communicate, she learned sign language and used pictures and was adding single words. She was polite – she could sign or say thank you when she had finished eating. It was still too early to assess how well she will eventually learn, but as a toddler she loved looking at books and newspapers, she understood the use of the remote control for the television and she knew that by the count of 10, she had finished her medicine.

Kim

Kim's birth in 1986 was difficult. She became distressed and needed to be delivered by emergency Caesarean. On delivery, she had breathing problems, her Apgar scores were low at 1, 5 and 6, she was light for dates and her hips were dislocated. Her first four weeks were spent in special care. Yet while Kim had one or two unusual features such as slightly pixie-like ears, a very small lower

Megan

Megan, 9 years old, has a duplication between 7q22 and 7q31. Apart from her mother's craving for jam and piccalilli sandwiches, the pregnancy was uneventful. Megan was born quickly and nothing unusual was noticed at the birth. Megan did, however, have congenital cataracts and wears glasses. Megan's difficulties with breast feeding were immediately obvious. She fed 'like a snapper turtle' and had swallowing difficulties and choked easily. Megan's ongoing difficulties with feeding resolved when lactose intolerance was diagnosed.

From birth, Megan had very low muscle tone. Her joints were so flexible that 'you could pack her into a very small bag'. On her back, she could lie totally flat, although her hip sockets were well formed. The low tone meant that Megan needed supports for her ankles and was late to crawl and walk. Despite this delay, by the age of nine, she was walking, running and swimming. As her muscle tone improved, a curve in her spine straightened. She has needed physiotherapy to make sure that her feet plant correctly to the floor. Her low muscle tone and double-jointed thumbs have affected her hand use but by the age of nine, she was able to use large-handled cutlery. She found holding a pen or marker difficult but was competent with a touch screen computer. At 9, she still needed help with dressing.

Megan's learning ability has been moderately affected by her chromosome disorder but her verbal skills were well in advance of her family's expectations. Her reading was ahead of her age group and in conversation she used language appropriately and well. She learned well in a group and 1:1 and once learnt, lessons stayed. At the age of nine, Megan was a 'very happy and lively' child. She had no sense of danger and needed tightly-drawn boundaries but with maturity her behaviour had grown increasingly controlled.

Aarie

Aarie, 10 years old, has a duplication between 7q22.2 and 7q32.2. At birth he was a good weight, but very floppy and had to be resuscitated. Breastfeeding was too demanding, so initially he was bottle fed but was repeatedly very sick with feeds until switched to a soy-based formula. At 10 years old, he was still taking blended foods and was able to spoon feed himself, sometimes with help. He had constipation and regularly took lactulose to control it. His growth and weight were average. Apart from repeated ear infections and frequent chest infections including pneumonia due to his generally low muscle tone, Aarie's health has been good, although he had hospital visits for surgery to correct a hernia and insertion of grommets to improve his hearing. By the age of 9 he had developed a significant sideways spinal curve which was surgically corrected.

Regarding development, Aarie had a high degree of dependency and needed close supervision. His continuing low muscle tone and loose joints made fine motor control difficult and delayed the achievement of his mobility milestones, so he rolled at 12 months, sat independently and crawled at 18 months and walked at three and a half years old. At 10, he was able to walk unsupported indoors but used a wheelchair for out of doors. Aarie communicated with gestures, vocal noises and pictures and while he had no speech, had very good comprehension. He has learned slowly and steadily and has been helped by his determination to learn. At 10 he could recognise numbers, enjoyed music, videos, books and basic touch screen computer games. He attended a special school and learned best in a 1:1 setting.

Duplications from 7q21-22 to the end of the chromosome

At the time of writing, *Unique* did not have any members with this duplication and the only reports were therefore those from the medical literature. This may mean that the picture painted looks rather bleak. From the medical literature, it appears that babies with this very large duplication do not generally do very well. Birth defects, some of them major, seem to be common and can affect survival.

Babies can expect to experience developmental delay. Among the babies described in the medical literature, birth defects have commonly affected the heart, brain and digestive system. More minor problems affecting the palate (cleft), feet (talipes, club foot) and spine (curvature) have also been seen (Muneer 1982; Forabosco 1988; Haslam 1992; Ishii 1997; Stetten 1997; Courtens 2001; Rodriguez 2002).

Duplications from 7q31 to the end of the chromosome

The information, almost exclusively from the medical literature, focuses on early problems rather than on development. It does appear that the rate of health problems in the first year of life is high. *Unique* has experience of one young child.

Common features, most of them found quite frequently in other children with chromosome disorders, include:

- Low birth weight
- Developmental delay
- Learning difficulties
- Growth delay
- Feeding difficulties, partly due to weak sucking
- Unusual brain structure
- Heart conditions
- Underdeveloped lungs
- Cleft palate

- Unusually large fontanelle (soft spot)
- Unusual features of the skeleton, for example, missing pair of ribs
- Specific facial features, including: low set ears, widely spaced eyes, small eye openings, small nose, small, receding lower jaw, long eyelashes
- Unusual muscle tone, either low so the baby is floppy, or high
(Alfi 1973; Vogel 1977; Novales 1982; Johnson 1986; Verma 1992; Strovel 1996; Boceno 1998; Courtens 2001; U)

Duplications from 7q32 to the end of the chromosome

Babies and children with this duplication, often linked with a deletion from another chromosome as part of an unbalanced chromosome translocation, appear to make somewhat better progress than those with a larger duplication extending into band 7q31. The oldest person described in the medical literature was 29 years old. However, almost all the information comes from people diagnosed with conventional cytogenetics rather than using a molecular technique, such as FISH (fluorescence in situ hybridisation) testing that might give more detailed information. The most common features described in the medical literature, many of them found frequently in children with other chromosome disorders, include:

- Low birth weight but an inconsistent growth pattern, with fewer than half the children described being short for their age
- Developmental delay
- Delay in mobility, which is typically quite marked. Not all children become mobile
- Some level of learning difficulty
- Low muscle tone, but high tone may occur in the lower body
- Skeletal anomalies, with some contracted joints, notably the fingers
- Kyphosis or scoliosis (forward or sideways spinal curve). This does not usually appear to be severe
- Congenital hip dislocation
- Heart problems
- Weak sucking and marked feeding difficulties
- Squint (strabismus), which can usually be corrected by patching or surgery
- Palate (roof of the mouth) is typically high and occasionally cleft (split)
- Unusual position of the feet

Unique's experience suggests that narrow ear canals and seizures may also be typical and that constipation is a frequent and troublesome complaint. Most children have very frequent lower respiratory tract infections and episodes of pneumonia and appear to be prone to reactive airways disease.

Puberty has been described twice, once in a girl with delayed and diminished signs