Deletions including the end of 13q

Inform Network Support

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Unique publishes three other leaflets on 13q deletions: 13q deletions including RB1; 13q various deletions; 13q distal interstitial deletions

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 Professor Dorothy Warburton, Professor of Clinical Genetics and Development, Columbia University, New York, US and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, UK 2006. Revised 07/2009.
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A 13q deletion is a rare genetic condition. It is caused by having lost material from one of the body’s 46 chromosomes. Generally speaking, losing chromosome material increases the risk for problems such as birth defects and growth and developmental delay. With 13q deletions the picture is very variable, depending mostly on how much material has been lost and from which part of the chromosome.

Chromosomes are the microscopically small structures in the nucleus of the body’s cells that carry genetic information. They come in different sizes and apart from the sex chromosomes (two Xs for a girl and an X and a Y for a boy), they are numbered 1 to 22, from largest to smallest approximately according to size. Each chromosome has a short (p) and a long (q) arm. In a 13q deletion, material has been lost from the long arm of one chromosome 13.

This leaflet describes the experience of people where the end of the chromosome has been lost. The chromosome may be simply broken, or the short and long arms may have joined to form a ring, or material from a different chromosome may be attached to the broken end of 13q.

How common are 13q deletions?

More than 140 people have been described in the medical literature and at the time of compiling the information in 2006, Unique had more than 100 members with a 13q deletion. Unique can put families who wish to make contact in touch with each other.

An additional observation has been made that Hirschsprung’s disease may occur in association with a 13q22 deletion. In this condition the nerve cells that normally control the rhythmic contractions that lead to matter being pushed through the intestines are missing (Kiss 1989). The kidneys may also be affected (Gershoni-Baruch 1996).

However, Unique has three members with this deletion: while all are disabled and significantly affected, they are making progress. One girl has feeding difficulties, hearing impairment and a developmental defect in her right eye that affects her vision, she is slowly gaining weight and at the age of 3 has started signing and making sounds and attends an opportunity playgroup. Another girl has good vision but a significant sensorineural hearing loss. She is unable to suck properly and is fed by gastrostomy tube. She has had successful surgeries for an encephalocele (protrusion of the brain through the scalp) and for a bowel obstruction. Two of the girls are known to have experienced premature puberty. Two also have a marked vulnerability to infection, and specifically to chest infections. A third girl with a small head and underdevelopment of the cerebellar vermis (the narrow worm-shaped structure between both sides of the cerebellum, the area at the back and bottom of the brain that plays an important role in movement and co-ordination) has no other apparent brain anomalies and at birth had normal vision. Although her development is delayed, she is communicative, expresses her feelings and plays. Her chief problems have been with weight gain and growth.

Breakpoint between 13q11 and 13q14.3

At the time of writing, Unique does not have any members with such a large deletion from 13q, but an 8-year-old child has been described in the medical literature with mosaicism for a 13q12 deletion. This means that some of her cells had normal chromosomes; others had a chromosome 13 with this large deletion. She had many features typical of a 13q deletion: her right fifth finger was missing and on the left side the finger was very short and her left thumb was also very short. She had a low hairline, bald patches on her scalp, and large ears with upturned lobes. She had a small encephalocele (a protrusion of brain tissue through the skull). At birth her hips were dislocated; however, she learned to walk, although used a wheelchair out of doors.

Although she had no speech, she was highly sociable, could communicate her likes and dislikes and had a boisterous sense of humour with people she knew well. At the age of 9, she developed seizures (Bamforth 1997).

An unborn baby has been described with mosaicism for a 13q13 deletion, following a mid-pregnancy scan that showed enlarged ventricles within the brain and a small encephalocele (protrusion of the brain through the scalp). The baby was also lacking a right eyeball, had a cleft palate, talipes (clubfoot), an enlarged heart and unusual genitalia with a phallus but no scrotum (Smith 2005).
fifth fingers (the metacarpals) may fuse, and the fifth finger or toe may be missing or very small. Specialised hand surgeons can reconstruct the hand to allow the index finger to do the job of the thumb (Brown 1993; Grindel 1999; Rodriguez de Alba 1999).

Hearing
The boy has a 70 decibel hearing loss and has very narrow ear canals. He wears hearing aids. One girl has normal hearing but in the other girl hearing is described as poor.

Vision
Vision is affected in all children. The eyes may be small, with small corneas. One girl has had detached retinas and there is a question mark over the boy’s ability to see: the retina and optic nerves are underdeveloped and he has a developmental defect in the structure of the eye. With vision therapy his eyesight continues to improve. The other girl is short-sighted and has had surgery to correct her strabismus (squint) (Wilson 1984; Karimi-Nejad 1998; U).

“J is the sweetest child or person I have ever met
“K gets frustrated at not being understood and bites, pulls hair and shouts

Food and eating
Unique’s members have had great difficulties with feeding and digestion. With a cleft palate, breastfeeding was impossible for the boy but he learned to drink from a bottle with a special Haberman teat. His appetite remained small and drinking thin liquids remained difficult so at the age of four he still had them thickened and ate bread and soft or pureed foods. One girl initially took breast milk from a bottle with an orthodontic teat but disliked drinking from an open cup and was fitted with a feeding tube (gastrostomy) direct into the stomach at the age of 6.

“ He enjoys playing with other people especially children. He likes music, swimming, snuggling and his special toys
“ She goes to Rainbows with 1:1 support. She likes music, TV, playing with mirror chimes and noisy toys and playing with other people

Breakpoint at q21 or q22
Unique’s experience is that the outlook for people with a breakpoint at 13q21 or 13q22 can be much better than is suggested in the medical literature. In addition to all the typical anomalies associated with smaller deletions from 13q, the literature suggests that in many people such a large deletion will have a marked effect on the development of the brain and the central nervous system. Effects on the structure of the brain may also be seen when the deletion is smaller, with a breakpoint at 13q31. The brain itself may be small and the forebrain may not separate fully into two hemispheres. The skull may have an open defect through which brain tissue protrudes or the vault of the skull may not form. There may be neural tube defects. Hydrocephalus may develop with excessive fluid in the ventricles. In the

Breakpoint at q34
People with a break at 13q34 have lost a relatively small amount of material from the end of the chromosome.

Reports in the medical literature show that the features of a small deletion with a breakpoint at 13q34 can be quite variable. There is likely to be some developmental delay but the degree is highly variable, ranging from none at all to severe.

Some people are completely healthy and look no different from other people. Others have specific traits and recognisable facial features, some of them typical of 13q deletions. Features that are typical of 13q deletions include a defect in the formation of the scalp leaving an area that is not a typical soft spot (fontanelle) uncovered by bone at birth, but which eventually covers over; bald patches on the scalp; a hole between the chambers on either side of the heart – atrial septal defect (ASD) or ventricular septal defect (VSD) – which may resolve naturally or be closed surgically; hypospadias (misplaced hole on the underside of the penis); low levels of certain blood chemicals, specifically coagulation factors VII and X, but not usually low enough to cause bruising or bleeding difficulties; and defects in the formation of single bones in the spine (hemivertebra), possibly leading to a spinal curvature that may be present at birth and may also progress. Other features described include microcephaly (small head) and lymphoedema (an abnormal collection of lymph fluid in the tissues, producing swelling) (Pfeiffer 1982; Flint 1995; Fryns 1995; Brewer 1999).

Unique currently has six members with a 13q34 deletion, two adults and four children, the oldest 33 years and the youngest two years old. Two have a simple deletion from band 13q34, the others have additional material from chromosome 3, 4, 12, or 20 added to the end of 13q which may affect them as much as the loss of material from chromosome 13. From the information available from parental reports, the pattern of common features is generally mild and varies from child to child. A hearing impairment was the only feature to affect two children, one of whom had very narrow auditory canals; other features affected one child each – a retinal defect known as combined hamartoma of the retina and retinal pigment epithelium; extra fingers and toes and an unusual foot shape; small genitals until puberty; small kidney; seizures; both inguinal and umbilical hernias; dislocated hips at birth; a squint (strabismus).

Size and growth
In Unique’s experience, babies were a similar size and weight at birth to babies without a chromosome disorder. The birth
From 1 to 4 years he seemed to put on weight quickly, from 4 to 14 he was average or slightly above; from 14 to 20 he put on weight rapidly. Weights of Unique members ranged from 2788g (6lb 2 oz) to 4195g (9lb 4oz). Growth was then steady and even the smallest baby at birth was tall for her age at 8 years. One of the adults is tall at 6'3" (190 cm) and both are large for their height.

Learning
This, the smallest loss of chromosome material from 13q, appears to have the most variable effect on learning ability. Here are snapshots of four Unique members:
- At age 2, performing at the level of a child nine months older
- At age 7, performing as a 5-year-old in all aspects of learning. She attends a mainstream school with a statement of special educational need
- At age 15, severe learning difficulties. She can recognise written names and can copy some writing
- At age 23, reads newspapers and magazines about subjects that interest him. Can write words with help with spelling, mostly uses the computer for letters. Has an excellent memory and a great sense of direction but poor concentration. After attending a special school to the age of 18, he moved to a vocational training centre to the age of 21 and now attends an adult day centre.

Speech and language
In general, the effects on speech and language mirror the effects on a child’s ability to learn. All the members of Unique on whom we have information talk, but speech in some is unclear and hard to understand.
- A 2-year-old is already talking
- A 7-year-old speaks in four or five-word sentences. She started to speak at the age of four. She sometimes has difficulty making herself clear and has to have things explained to her many times.
- A 16-year-old speaks and is understood by her family. She finds it frustrating that people she doesn’t know cannot understand her.
- A 23-year-old communicates well and is easily understood. His speech pattern was good from the age of 2 although his understanding trails behind his speech. He has good pronunciation.

He asks long and complex questions and sometimes doesn’t wait for the reply to the previous question. He sometimes can’t find the right words but generally speaking can express himself.

Sitting, standing, moving
The evidence from Unique is that babies are only mildly delayed in reaching their developmental milestones for movement. Unique babies rolled over for the first time between 6 and 9 months; sat unsupported between 9 and 12 months; and took their first steps in their second year. One child has shown evidence of balance and co-ordination problems once she was walking.

Size and growth
The Unique children were a normal weight at birth – between seven pounds (3175 g) and almost ten pounds (4500 g). Children are not especially short or receiving treatment to boost their height. By age 4, the parents of the boy believe he is 10 to 15 per cent shorter than he would be with regular chromosomes. One girl is tall and lanky and has only had a problem putting on weight.

Learning
Children are good at recognising and remembering people although they may not communicate this at the time. One attends a mainstream pre-school with 1:1 support; the other is in special education; the third has completed education. Their level of learning disability is described as profound.

- His love of life helps him to learn
- Being stubborn and determined helps her to learn

Speech and language
Children communicate with vocal noises, facial expressions and gestures as well as a signing system. At 7, one girl uses single words and a one-step communicator switch. Her mother says that she gets very frustrated by her inability to make the sounds of speech clearly.

- There is a huge difference between what she understands (90%) and what she says
- He understands quite a lot more than we even presume

Sitting, standing, moving
By 19 months, the boy was able to roll from front to back and by two he was starting to use a standing table. One girl could sit by the age of three and by seven was commando crawling to reach her toys and walking in a walker with trunk and pelvic support.

Medical concerns
Two children had a heart condition at birth: the boy had a persistent ductus arteriosus that was closed surgically when he was two years old. He also had abnormal heart valves and a hole between the upper chambers of the heart. The girl had holes between both upper and lower heart chambers and had an operation to close them in her second year. The boy had hydrocephalus but this did not need treatment. Both children developed more respiratory infections than other children or are sicker when they catch them. The girl also has multiple allergies. The girl wears a body jacket to control her hydrocephalus, but this did not need treatment. Both children developed more respiratory infections than other children or are sicker when they catch them. The girl also has multiple allergies. The girl wears a body jacket to control her spinal curvature (scoliosis). She wears toe separators to straighten her overlapping toes and supports to prevent her feet from rotating inwards. Dislocated hips and talipes (club foot) have required surgery.

Hands and feet
Underdevelopment or even absence of the thumbs or less commonly the big toes can occur when band 13q32 has been lost, suggesting that a gene or genes important for the development of thumbs and toes is to be found in band 13q32. Less commonly, the fifth finger, especially the middle joint, can be unusually short and often there is a single crease in the finger. Occasionally the bones that lead from the wrist to the fourth and fifth finger, especially the middle joint, can be unusually short and often there is a single crease in the finger. Occasionally the bones that lead from the wrist to the fourth and
Children were typically born with a small head, although by age 4 the size was in proportion to the rest of the body. In one girl, the skull bones fused early.

Medical concerns
One child has one kidney that is smaller than the other. This does not cause problems but she is monitored regularly. This same child also tends to develop asthma when she has a cold. One of the adults had small genitals until puberty. The adult size is normal.

Two children bruise easily. Both have a simple deletion from the end of the chromosome with no additional chromosome material.

One of the adults has very large and unusually shaped feet with a small big toe. He was born with an extra toe on each foot and an extra finger on each hand.

Hearing
Two children have a hearing impairment. One has a permanent loss in her left ear and wears hearing aids; the other wore a hearing aid but at 16 no longer needs one.

Vision
One child with a simple 13q34 deletion has a condition known as combined hamartoma of the retina and retinal pigment epithelium, in which the retina is wrinkled and contains tortuous blood vessels. Treatment with cryotherapy may prevent haemorrhage.

Behaviour
The Unique series is not large enough to suggest any consistent effect on behaviour. Three snapshots follow.

- At 7, very confident and has very challenging behaviour. Can be quite loud and can push and shove a lot
- At 16, generally a very happy child, affectionate and demonstrative. Likes routine, needs to feel secure and needs lots of reassurance.
- Challenging behaviour first manifested itself from 14. At 23, he had calmed down a lot and only has an occasional outburst when he doesn’t get his own way. He never had a formal diagnosis of autism, but he does have autistic tendencies. He can be moody and challenging, but can also be very helpful.

Independency?
Unique has no reports of adults living independently. The 23-year-old lives with his parents in a tight-knit and supportive community to which he is able to contribute. He works at an adult day centre where he is paid a small sum to complete contract work. He is able to shop independently at a supermarket and to use public transport on his own. He doesn’t drive. Every third weekend he spends in a group home as a respite facility. He enjoys swimming and basketball and belongs to clubs for them. His parents say that he is very good with his nephews and nieces and says he wants to marry.

Breakpoint at q33
The effects of a loss from band 13q33 depend at least in part on the breakpoint. FISH or other molecular genetic studies will indicate this and your geneticist will explain it. However, a straightforward link between the breakpoint and the effects of the chromosome loss cannot be made, although in general, the larger the amount of band 13q33 that is lost, the greater the effects. The only reasonably consistent effect is some
degree of developmental delay. What follows is a list of the common effects known from a study of the medical literature. However, not every child is affected; some may have only minimal effects; others may show one or two typical effects; in others, more organs are involved. Unique’s experience is that its members are often more mildly affected than the medical literature suggests.

The body systems that are typically involved with this breakpoint include the spine, where individual vertebrae may not be fully formed; the hands, where the fifth fingers may be unusually short; the anus (bottom), which may be covered over; the genitals may show a variety of anomalies; the kidneys; the heart; the head, which is typically small (microcephaly); the ears, typically large, with fleshy lobes; and the eyes, which may show a structural or developmental defect, such as a coloboma (showing as a ‘keyhole’ iris) (Turleau 1978; Mucke 1983; Stoll 1998; Kuhnle 2000; Luo 2000).

Unique currently has six members with a breakpoint at q33, four with a simple deletion from band q33, q33.2 or q33.3 and one with a complex karyotype involving additional material from chromosome 8. From the information available from parental reports, every child has unusually frequent infections, three have a small head, three have a hearing impairment, two have a kidney or urological concern and two have a squint (strabismus). One child had a persistent ductus arteriosus (a persisting fetal organisation of blood flow from the heart that sometimes corrects itself but may need surgery); an inguinal hernia and a stricture affecting the spleen: both were corrected surgically. Medical concerns that are described in the medical literature but occur in only one child within Unique include frequent bruising and copious bleeding (see breakpoint at 13q34) and unusual position of the anus close to the genitalia (U).

A child with a breakpoint at 13q33.3 is described in the medical literature as having growth delay but his ability to learn was not severely affected. He has a small head, hypotonia (low muscle tone) and slightly unusual facial features including large and low set ears, a low nasal bridge, widely spaced eyes, a small chin and a high, broad forehead (Stoll 1998). Unique has a member with this deletion: his key features are ambiguous genitalia, severe conductive hearing loss, urinary tract infections, vulnerability to infection, squint, small head, bruises easily and bleeds copiously, rectum sited beneath testicles; he also has hypotonia and an autistic spectrum disorder (U).

Three boys with a breakpoint at 13q33.2 or 13q33 have been described in the medical literature. One had a neural tube defect (a myelomingingocele) in addition to a small head, widely spaced eyes and ambiguous genitalia. He also had a neurogenic bladder (loss of normal bladder function caused by damage to part of the nervous system), undescended testicles, mild scoliosis, mild hydrocephalus, developmental delay and marked learning difficulties. The other boy also had unusual genitals, with a small penis placed beneath the scrotum and the hole usually at the end of the penis on the

### Speech and language
The variation of ability in learning is also seen in children’s ability to speak. Unique records suggest that more able children learn to speak and acquire a vocabulary of single words that alongside signing and gestures allow them to make their needs and wishes understood. Less able children are not able to communicate at this level.

### Sitting, standing, moving
Unique records suggest that babies are particularly delayed in reaching their ‘milestones’. The earliest age at which a baby sat alone was seven months, the latest was 24 months, but at least one child was not able to sit at the age of five. One boy was said to be walking by the age of two, but most parents said their child took much longer than this. By the age of four or five some children are walking with the help of a walker and splints but will still need a wheelchair for anything other than short distances. By contrast one girl’s favourite activity is football.

### Medical concerns
- **Hips**
  Developmental disorders of the hips have affected five out of Unique’s nine members. One boy has osteoporosis and acquired hip dysplasia, two have needed surgery to enable them to walk and two others are currently being monitored.

- **Heart conditions**
  Heart conditions also affected five children but resolved naturally so that surgery was only needed for one child. Three children had a septal defect – a hole between the chambers of the heart - which closed over naturally and one child had a heart murmur that turned out to be benign. One child had a persistent ductus arteriosus that was closed surgically when she was four. She also had a bicuspid aortic valve consisting of two leaflets instead of three. One baby had a vein of Galen malformation in which the capillaries connecting the arteries with the veins in the brain are missing, leading to increased blood flow and increasing the strain on the heart.

- **Kidney conditions**
  Two babies were born with unusually small kidneys and by the age of nine they were beginning to lose function in one child. One child was born with a single kidney.

- **Respiratory infections**
  Respiratory infections are common in babies and children with a tendency to bring back some of their feeds and to aspirate them. Three older children in this group aged between six and 12 are also noted to have a marked tendency to severe asthma.

### Other conditions
Other conditions affected individual children only. They included pyloric stenosis, seizures, scoliosis, deficiency of blood clotting factor VII, osteoporosis, cleft palate, fusion of ribs at the sternum.

- **Hirschsprung’s disease**
  This severe form of constipation caused by absence of nerve cells in part of the bowel has very occasionally been found in children with a 13q deletion but it is not certain whether it is part of the syndrome or incidental to it (Bottani 1991).
Breakpoint at 13q32.3
Three people have been described in the medical literature including a 49-year-old man, with few features in common. One baby was born prematurely at 33 weeks after a pregnancy in which there was very little amniotic fluid; he was small for dates and had unusual genital features including a divided scrotum and very small penis; he later developed severe feeding difficulties and gastro oesophageal reflux. His development was delayed and he took his first steps and said his first words at 2 years, although his speech was particularly delayed and his developmental quotient was judged to be 50. A second baby was markedly delayed but had no major physical defects. The adult man had uneven sized pupils, developmental delay and equinovarus (clubfoot) of the left foot, corrected with surgery. He was short. Three vertebrae were fused (Rivera 1985; Uriostie 1995; Van Buggenhout 1999).

Among Unique members, three have this deletion, one together with a duplication of material from chromosome 12. According to family reports, all have experienced some delay in motor development and ability to learn and specifically in speech. One child has developmental dysplasia of the hip, another has a cleft palate and a missing heart valve; two have problems with eyesight and one has impaired hearing. In terms of learning, a 12-year-old with severe learning difficulties can copy and write but cannot read; she can use a computer and has a good memory. Her single-word speech is developing steadily (U).

Size and growth
Babies are usually small at birth. Among Unique babies, birth weights ranged from 2126 grams (4lb 1 oz) to 3401 g (7lb 8oz). Growth is then typically delayed and some children are remarkably small. One baby of nine months was still wearing newborn clothes and three families specifically described their older children as unusually short. At the age of 12, one girl is under four foot tall. Although his growth hormone levels were normal, one boy took growth hormone for a year to boost his eventual height.

Food and eating
Unique’s small series does not show a distinct pattern of feeding difficulties that would set affected children apart from other children with chromosome disorders. At least one child breastfed well and has continued to eat well to her present age of 11. Two children are reported to have developed gastro oesophageal reflux. This can usually be controlled by feeding with thickened drinks and careful positioning during and after feeds. If the reflux is a continuing problem prescribed antacid medications will protect the food pipe. However one child has been fed by gastrostomy direct into the stomach.

Learning
Unique records show a variable level of learning difficulty. At the age of 5, one girl attends a mainstream school with 1:1 support, is working hard to write hand over hand and doing reading practice. Another girl of the same age is achieving at a much lower level. An 11-year-old can draw letters, numbers and faces, copy words and write her own name. She started to write at the age of 7 and looks at books but cannot read. At 10, she started to use a computer and is reported to have a very good memory. The overall learning disability of these girls would be described as moderate to profound.

Speech and language
At three, a girl understands both Spanish and English but is not yet using words. She points, cries and signs instead. She is awaiting a formal diagnosis of severe apraxia.
- At four, a boy started to use words. Now aged eight, he mingles words with vocal noises and gestures. He usually speaks in three-word sentences but loves long and complex words. He has particular difficulty in making the sounds of speech clear.
- At 6, a boy uses single words and sometimes links them, for example: thank you. He understands well and is frustrated at his inability to express himself. His lack of tongue control makes it hard for him to speak clearly.
- At 12, a boy speaks, but not clearly enough to be understood outside the family.

Sitting, standing, moving
Delay in reaching gross motor milestones is a particular feature. Unique’s information shows babies rolling over from 10 months, sitting around the middle of their second year, bottom shuffling or crawling a few months later than this and walking between two and a half and three and a half. This picture suggests a uniformity that is not the case. A three-year-old is now active and surprising her doctors by her ability to run, while the eight year old boy needs cushions to sit and supports, adapted footwear and a walker in order to be mobile. A six-year-old walked from two and a half, but he is unsteady, tires easily, wears a leg strap to correct the angle of his legs, and needs regular physiotherapy.

Medical concerns
- Infections
Infections are both frequent than expected and relatively severe. Two children are reported to catch frequent chest infections and need hospital treatment. Another is reported to catch virus after virus and a fourth is said to have low immunity. In one child a connection was found between the oesophagus (food pipe) and the airway (Walsh 2001).
- Kidneys
One child was frequently found to have traces of blood in his urine as a baby, suggesting longstanding urinary infections. Another has small kidneys and has severe reflux from the bladder to the kidneys. This was corrected surgically.
- Unusual genital features
A gene, probably sited at 13q33.2-q34, is thought to regulate the normal development of the genital area. Some children in whom this gene is missing develop unusual genital features. Boys and girls can be affected, but the problems are generally more obvious in boys. Within the Unique series, twice as many children were not affected as were affected. There is a range of defects that includes hypospadias (the hole usually at the end of the penis is sited under the shaft); a very small penis; divided scrotum; and transposition of the penis so that it is sited beneath the scrotum. In girls the abnormalities include a divided uterus, an enlarged clitoris and absent labial folds outside the vagina. The anus (hole for the bottom) may also not be apparent at birth, usually because it is covered by skin, but sometimes because the rectum has not fully developed. Instead, a channel may lead from the rectum to the vagina in girls or to another opening near the scrotum in boys. Sometimes the anus develops normally but is very close to the scrotum (Urioste 1995; Bartsch 1996; Kuhnle 2000; Walsh 2001; U).

Other medical concerns that arose individually were a curved spine (two children, neither requiring surgery), inwards rotating feet and severe eczema.

Hearing
Two children have worn aids for a hearing impairment, but one child no longer needs them at age 6. One of these children has also had grommets (ear tubes) fitted and one has had repeated ear infections, damaging the ear drum. By contrast, another child is described as over sensitive to sound.

Vision
Apart from one child with an astigmatism, none of the children in the Unique series has a structural eye defect but two have strabismus (a squint) and one is possibly very short-sighted although this has not been formally diagnosed.

Breakpoint at q32
“A great giggle and a pretty even temper
Rachel laughs and sings all the time and makes you so happy to be around her. She is a very bright child and will attempt most things - age 12

There is considerable individual variation between people with a breakpoint at 13q32. It has been suggested that involvement of band 13q32 in a deletion implies more extensive and severe disability, but Unique records suggest that this is not necessarily the case. Descriptions from the medical literature show that features can include, in addition to those seen with breakpoints at 13q33 or 13q34: small size following slow growth after birth; a small and unusually shaped head, sometimes with a low hairline over the forehead or coarse patches of hair on the scalp; underdevelopment of the band of nervous tissue that connects the two hemispheres of the brain (agenesis of the corpus callosum) or other structural anomalies of the brain; increased hair growth over the body; a hearing impairment; underdeveloped or missing thumbs and sometimes other unusual features of the hands or feet; a heart condition; eye conditions including absence of the iris; scoliosis and talipes (club foot) (Telfer 1980; Wilson 1984; Al-Awadi 1985; Brown 1993; Grindle 1999).

Unique currently has nine members with a breakpoint designated at 13q32, aged from three to fourteen. Of these, six children have a breakpoint designated 13q32, and in two of them another chromosome is involved, either with material missing or duplicated. Three of the nine have a breakpoint designated 13q32.3. From the information available from family reports, a pattern of common features emerges. Heart conditions and disorders of hip development affect five children out of the total of nine; vision impairment and short stature affect four children; and a small or unusually shaped head, kidney conditions, a marked tendency to asthma and hearing impairment affect three children.

“He would take his hearing aids out and chew them and then lose them and it didn’t make very much difference. His hearing has now improved so he no longer needs to wear aids unless he has a prolonged cold.