

Unique

Ring 13



Ring 13

Ring chromosome 13 is a rare genetic condition caused by having an abnormal chromosome 13 that forms a ring.

What is a chromosome?

All our genetic information is contained in the cells of our body. The genetic material that contains this information is the DNA, tightly coiled and forming a number of rod-like structures called chromosomes. Genes are fragments of the DNA strand, which contain coded instructions for the formation of different proteins that control most tasks in our body. Genes constitute our genetic blueprint, and there are 20-25,000 genes located on 46 chromosomes. These 46 chromosomes occur as 23 pairs. We get one of each pair from our mother in the egg, and one of each pair from our father in the sperm. The first 22 pairs are numbered 1 to 22, approximately from longest to shortest. The remaining pair are the sex chromosomes. Girls and women usually have two X chromosomes (XX) and boys and men usually have an X and a Y chromosome (XY). Each chromosome has a short arm (p for French petit) and a long arm (q), separated by a pinched area known as the **centromere**.

Chromosome 13, along with chromosomes 14, 15, 21 and 22, has a distinct shape with a very small short arm that does not contain genes that are relevant to development. This group of chromosomes is known as **acrocentric** chromosomes, meaning that the centromere is near one end.

Development works with clockwork precision and the right amount of genetic material is needed for normal growth and development. If there is a substantial amount of missing or extra genetic material, then it is likely to result in problems in growth and development and most often also in the functioning of the body systems.

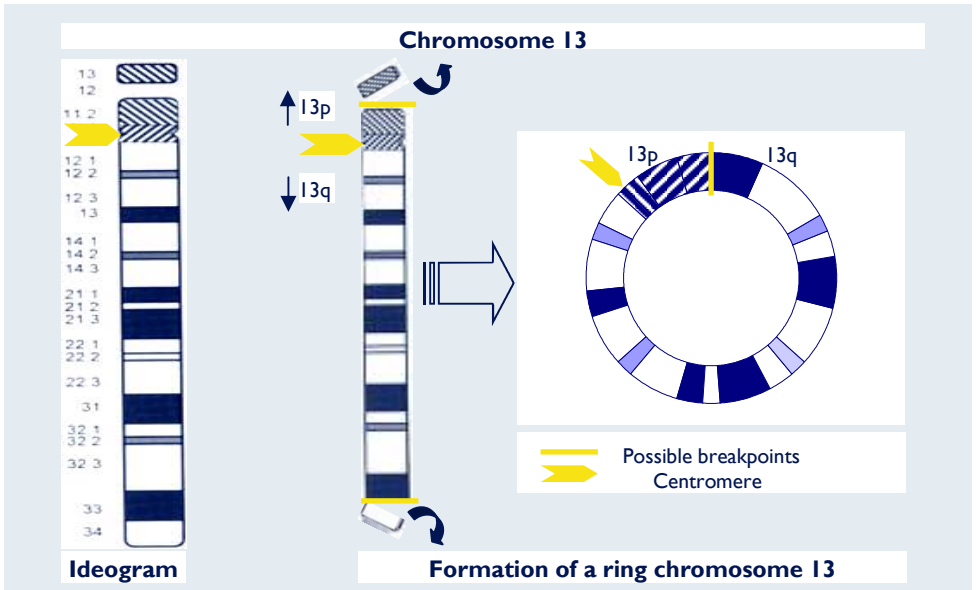
References

The text contains references to articles published in the medical press. You can search for the abstracts or original articles on the internet in PubMed. References to information from Unique are marked U.

What happens when a ring chromosome forms?

In people with ring chromosome 13, one chromosome 13 is usually intact but the other has formed a ring. When a ring forms, both arms of a chromosome break and the broken 'sticky' ends fuse at the breakage points. The broken fragments are lost, and with them any genes they may contain. In the case of a ring chromosome 13, only the genes in the long arm (13q, at the bottom of the chromosome in the diagram above right) matter.

Ring chromosomes also have particular effects on cell growth. During development, cells naturally divide, but the ring chromosome formation may disrupt this process because the ring chromosome may not behave properly and become entangled, broken or double in size during cell division. As a result of this, cells may arise with the wrong amount of chromosome 13 material (too much or too little). This is called **mosaicism** and has a strong influence on the effects of the ring 13. Most commonly, people also have cells with 46 normal chromosomes, which generally softens the impact of the ring 13. The size of the ring chromosome may also vary between different cells and different tissues of the body.



How chromosomes are studied

In a routine chromosome analysis the white blood cells are cultured for analysis. When these cells are about to divide the chromosomes become more compact and can be visualised under a microscope. The cell is then pre-treated and stained with a dye to allow a specific pattern of different-sized light and dark bands to be seen when they are viewed at up to about 1000 times life size under a light microscope. These bands are numbered according to an international system. A diagram that represents the banding pattern of a given chromosome is called an **ideogram**, which is the equivalent of a map of that chromosome. Careful analysis determines if the chromosomes are complete or if there is extra or missing material.

Why did this happen?

The great majority - 99% - of ring chromosomes are sporadic. The actual cause is not known and should be regarded as an accident that happened in cell division in the process of making sperm or egg cells. These accidents are not uncommon. They affect children 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 ring to form.

Very occasionally, a ring chromosome may be inherited from a parent. In most (90%) of these familial cases the ring has been inherited from the mother, as ring chromosomes appear to be associated with reduced fertility in men. To date only one mother-daughter pair with a ring chromosome 13 has been reported in the medical literature and both are very mildly affected (Bedoyan 2004).

Will it happen again?

So long as tests show that parents' chromosomes are normal, they are very unlikely to have another affected child. All the same, you should have a chance to discuss prenatal diagnosis if you would like it for reassurance.

What are the medical consequences of any ring chromosome?

The process of disruption described on page 2 results in what is known as **ring chromosome syndrome**, regardless of which chromosome is involved.

The most common features of ring chromosome syndrome are:

- Slow growth and eventual short height
- Some level of learning difficulty or disability
- Very small head (microcephaly)
- Some unusual facial features. These can be subtle or more obvious
- Anomalies in skin colouring, such as streaky pigmentation, as a result of mosaicism

Does it help to know the exact breakpoints in the ring?

It can help to predict the severity of problems. If the breakpoint is closer to the end of the long arm (eg at 13q34 in the ideogram on page 3), a smaller amount of material and genes will be lost, and this will have fewer medical consequences. If the breakpoint lies higher up (eg 13q22), more chromosome material will be lost (nearly a third of the long arm), which will have more far-reaching medical consequences. The effects of the ring are also changed to some extent by any cells with a different chromosome make-up. Because of mosaicism, the findings in blood cells may not reflect precisely the chromosome make-up in cells of other body tissues and this makes it more difficult to make accurate predictions for a particular child.

Main features of ring 13 syndrome

In addition to the general features of ring chromosome syndrome, the most common features in ring 13 syndrome are:

- Developmental delay
- Feeding difficulties as a baby
- Unusual formation and position of one or both feet and sometimes toes
- High palate (roof of the mouth). More unusually, part of it may be cleft (split)

Some features occur more commonly in people with a break near the centromere.

- Unusual hands. Some fingers, particularly the fifth finger and specifically the middle joint, may be short. Thumbs may be unusually formed, in an unusual position and small or underdeveloped
- Unusual position or formation of the anus (bottom). It may be covered by skin, very small or unformed. This can usually be corrected by surgery
- Incorrect position for outlets to the urinary system. This can usually be corrected with surgery
- Some degree of genital abnormality. This may be very subtle and minor or more problematic
- Small lower jaw
- Kidney anomalies
- Heart defects
- Abnormal brain structures
- Unusual bone structure, most commonly rib fusion. This has only been reported in babies with a breakpoint near the centromere
- Abnormal eye structure, most obviously very small eyes. This usually affects vision

Features in ring 13 syndrome

■ **Very small head**

Most children have a very small head and the bones of the skull may be small compared with the size of the face. In a few children, some joints (sutures) between the plates of the skull fuse early, giving the head an unusual shape. In other children, the main soft spot on top of the head (anterior fontanelle) is small at birth and closes early. A small head does not need treatment. If it is lopsided or has an unusual shape because of early fusion of the skull plates, it is possible to re-open the sutures, but in children with ring 13 syndrome this has not been shown to help (Fried 1975; Jones 1981).

■ **Brain structure**

Babies with small ring 13s, where genes are missing from band 13q32, have a risk of brain defects. For this reason, it is usual for babies with known ring 13 to have a brain scan. This may be possible through the fontanelle. The range and severity of defects is quite broad, and includes underdevelopment or absence of the corpus callosum (the band of nerve fibres linking the two sides of the brain), failure of the forebrain and sometimes the skull covering it to develop, protrusion of part of the brain through a gap in the skull, defects of the cerebellum, the part of the brain that controls balance and co-ordination, enlarged ventricles and hydrocephalus. Where the brain or the forebrain has failed to develop, the bony covering may also be missing.

■ **Facial features**

The formation of the head and face is a complex process involving many genes. Everyone has some minor differences that are considered normal, such as large ears, a small nose or eyes close together. Whenever there is a significant loss of chromosome material and genes, there may well be more minor differences (unusual features) than one would expect in someone without a chromosome disorder.

In babies and children with ring 13, these typically include a wide bridge to the nose. The forehead may slope back to the hairline and the ears are very often large and may have an unusual shape or be set low on the head. Around half of all children with ring 13 have a noticeably small jaw and chin. Eyes are often set far apart and may have tiny folds of skin across the inside corners. Occasionally, children have hooded upper eyelids (ptosis). If the lid interferes with vision, it can be lifted in a small surgical operation. Other features remarked on by one or two families include a thrusting tongue and protruding teeth, slanting eyes and a skinfold on the forehead. In one child with a mosaic constitution, one eye may blink faster than the other (Magenis 1976; Lagergren 1980; Jones 1981; Martin 1982; Venugopalan 2001; U).

■ **Hair patches**

An occasional feature of ring 13 is patchy hair loss. This is not permanent, as the hair grows back (Verma 1978; U).

■ **Skin pigmentation**

Dark or light streaks, whorls or patches of skin occasionally occur. They have been described on the chin, the trunk and the back. In one child, the colouring faded by the age of 9 (Verma 1978; Steinbach 1981; Fryns 1998; U).



Loss of vision in left eye
Ring with breakpoint at 13q21.2

■ Eyes and vision

Children with a large ring with a breakpoint at 13q34 do not usually have serious vision problems. Minor problems such as long or short sight or a squint (strabismus) are, however, common and can be corrected with glasses, or in the case of strabismus sometimes with patching. Children who have lost a larger part of chromosome 13 do have a risk of having a structural eye defect. These have included a very small eyeball, underdevelopment of the optic nerves, cataracts, other defects of the cornea at the

front of the eyeball and coloboma, a developmental defect which may affect vision, depending on the part of the eye affected. Keratoconus has also occurred, in which the cornea viewed from the side is shaped like a cone, distorting vision. Children in whom band 13q14 has been lost are at risk of retinoblastoma, a rare form of eye cancer (Fried 1975; Jones 1981; Filous 1998; Heaven 2000; U).

“A had five sets of grommets fitted by the time he was nine years old as he had recurring severe glue ear. On the fifth occasion, the surgeon was only able to fit one grommet and it was decided to fit a hearing aid to his left ear. He has adapted amazingly well. He chose a blue ear fitting and was pleased to find elephant stickers to put on the part that goes behind the ear. When his right grommet comes out he will have another hearing aid fitted. A has always loved music, and since he has had his hearing aid has really enjoyed listening to his CDs. His speech has come on a great deal and he now says clearly words he could not say well before. It’s a joy to see the improvement and it means no more operations, which were always distressing for him and us.”

■ Ears and hearing

Hearing loss is not considered to be part of ring 13 and some children with a large ring and a small loss of material will have normal hearing. However, permanent hearing loss in both ears has been found in a child with a breakpoint at 13q14 and a mosaic constitution and a partial hearing impairment usually requiring hearing aids has been found in six out of 10 *Unique* children and adults. Hearing loss is also part of trisomy 13 (having an extra chromosome 13). One factor is that having a small head, children have narrow ear canals and this makes glue ear more frequent and more severe. In one child, no ear canals were found (Jones 1981; Goldsmith 1993; Gentile 1999; U).

■ Palate

Around half of children with ring 13 have a high palate (roof of the mouth). A high palate is common in children with a chromosome disorder and is caused in part by abnormal mouth and tongue movements

during fetal development. The high palate usually causes few difficulties, but it may make feeding more difficult. A cleft (split) in the palate is much less common but has been seen in two *Unique* members and two children described in the medical literature, one of whom also had a cleft lip. If you are feeding a baby with a cleft palate, you should be

offered specialist support and aids to help the baby swallow correctly. The cleft can be repaired surgically later (Jones 1981; Talvik 2000; U).

■ **Heart**

While not formally belonging to ring 13, structural heart anomalies occur more often than in children without chromosome disorders. They are much more likely to occur in children with a large deletion and a small ring than in children with a 13q32 or 13q34 breakpoint. The most common types of heart problem found at birth are holes between the upper or lower chambers of the heart (atrial or ventricular septal defects, ASD/VSD), persistent ductus arteriosus (PDA), which is a remaining structure from the fetal circulation and narrow valves between different parts of the heart or between the heart and a blood vessel. Some ASDs and VSDs close naturally but if they do not, they can be repaired surgically and a PDA can also be closed if necessary (Benn 1983; Gentile 1999; Lorentz 2002; Guala 1997; Talvik 2000; U).

■ **Ribs**

Babies and children with a large ring with a 13q34 breakpoint are most likely to have a normally formed ribcage with 12 pairs of separate ribs. In babies who have lost more chromosome material, it is common to see pairs of ribs fused together and extra pairs of ribs and in one baby the breastbone (sternum) had not formed. These are individual differences and do not need usually treatment (Martin 1982; Guala 1997; Gentile 1999; Lorentz 2002; U).

■ **Kidneys**

Most babies will have healthy functioning kidneys, whatever the size of their ring. However, a small minority will either have a single kidney, small kidneys or another developmental abnormality of the kidneys. These kidney anomalies can usually be managed with medication or regular monitoring (Hevia 1979; Martin 1982; Benn 1983; Lorentz 2002; U).

■ **Diabetes**

A small number of young people with ring 13 develop either type I or type II diabetes and need daily treatment with insulin injections. The known age range at which diabetes has developed is 12 to 23 years (Lagergren 1980; U).

■ **Intestinal malrotation**

This is a developmental anomaly of the digestive tract that can lead to intestinal blockage. It is unusual in ring 13 syndrome but has occurred in babies with a small ring and a large deletion (Benn 1983; U).

■ **Abnormalities of the anus**

These are seen commonly in babies with ring 13, particularly where the ring is small and the deletion is large. The anus may be imperforate, so there is no visible hole. The rectum ends in a blind pouch, which may be high up or low down, close to the skin.

An imperforate anus can be repaired surgically soon after birth. A low lesion can be repaired with a surgical operation known as a perineal anoplasty. Babies with a high lesion may first have a colostomy (an artificial outlet from the intestines), with later surgery to reconstruct the anus. In a few babies, the anus is small or is entirely normal, but positioned unusually close to the genitals (Brandt 1992).

“ Our daughter had no vaginal opening but all her internal organs were normal. We have opened up the vagina gradually when changing her.

■ Genital area

Many babies are born with some degree of genital abnormality. This is reported more commonly in boys but girls can be affected as well. Many of the abnormalities are the result of developmental defects in the division of the lower end of the intestines, leading to the rectum and anus, from the urinary and genital systems. There is a broad range of severity,

from babies with a common outlet (known as cloaca), to boys with a small penis, undescended testicles, a divided (bifid), small scrotum, hypospadias (where the hole normally positioned at the end of the penis is on the underside), ambiguous genitalia or a malpositioned urinary outlet. In girls, a common outlet and fistulas (connection) between the rectum and vagina have been seen, as well as failure of the vagina to open. If needed, most problems can be corrected with surgery (Magenis 1976; Niebuhr 1977; Benn 1983; Brandt 1992; Bartsch 1996; Boduroglu 1998; Talvik 2000; U).

■ Spine

A dimple or pit at the base of the spine has been observed in a few children, but this has not caused any problems other than the need to be particularly careful with nappy/ diaper cleanliness (Lorentz 2002; U).

■ Hands, fingers and thumbs

There is often something unusual about the hands, fingers or thumbs of children with ring 13, although this is less common in children with a large ring with only a small loss of chromosome material. Some of the more typical findings in children with small rings with a breakpoint around 13q22 are thumbs that are underdeveloped, missing or malpositioned or cannot be properly bent. The supporting bone structure in the hand (first ray) may also be absent. The bones in the hand that lead from the wrist to the third and fourth or fourth and fifth fingers are sometimes fused. The fifth finger and sometimes the first and second fingers and specifically the middle joint are sometimes short and occasionally fingers have been described as ‘spindle-like’. The fingers may be bent and overlap each other (Fried 1975; Schmid 1975; Magenis 1976; Steinbach 1981; Martin 1982; Mules 1983; Amor 2005; U).



13 months old

■ Hips

The hips may be unstable at birth and may be able to be dislocated. This is especially apparent in babies with a small ring and a large deletion. Treatment for dislocated hips may include physiotherapy and in some cases temporary immobilisation in a brace or cast and surgery.

■ Feet and toes

Between half and two-thirds of babies with ring 13 are born with abnormally positioned feet, toes or both. It is highly typical for one foot or both to be turned inwards, although this is seen less often in babies with breakpoints near the end of the chromosome at 13q32 or 13q34. In babies

with small rings, some toes also commonly overlap each other or are joined by skin. The fifth toe has occasionally been found to be very small and in two families where the child had a 13q34 breakpoint, the feet were described as unusually broad. Most foot problems can be treated with physiotherapy or splinting and if necessary a surgical procedure (Uccelatore 1990; Brandt 1992; Lorentz 2002; U).

■ Other concerns

Many other conditions have been described occasionally in babies and children with ring 13. These features may be a part of the specific chromosome disorder but they may also have occurred by coincidence. Some of these conditions include a restriction in the upper airways (trachea) and a cleft in the throat; coeliac disease; seizures; abnormal gallbladder; missing kneecap. Theoretically, the absence of the genes which code for blood factors VII and X could lead to bleeding disorders, but this has not been observed (Parcheta 1985; Brandt 1992; Talvik 2000; Lorentz 2002; U).

First signs

Some features of ring chromosome syndrome such as a baby's unusually small head and slow growth rate would normally be detectable during pregnancy. In many cases, the baby's slow growth was noticed during pregnancy (see below) but of 11 *Unique* families who gave information, only one baby, with a deletion from 13q21, was diagnosed before birth. For three babies the key area of concern at birth was their small head, and in one it was low birth weight. In two babies, feeding problems raised concerns and one baby failed to gain weight by the six-week postnatal check. Two families noticed that their baby was not developing normally – one baby was still unusually floppy at four months and another showed little interest in toys at six months. A girl with a mosaic chromosome make-up was diagnosed at school age, when speech delay was noticed (U).



Pregnancy

Among eleven *Unique* families who gave information on pregnancy, six babies were monitored closely because of their slow growth in the womb and three mothers were admitted to hospital because of this slow growth. One baby with a large deletion was the size of a 13-week fetus at 20 weeks. Two babies were induced before term at 37 weeks because of their slow growth rate. Some mothers commented on the lack of fetal movement, especially in the last three months of pregnancy. Otherwise, most mothers described their pregnancies as 'excellent' or 'normal.' Taken with information from the medical literature, the *Unique* series shows that babies with a small ring with a breakpoint at or above 13q32 were born on average between 36 and 37 weeks, while babies with a breakpoint at 13q34 were born on average just before term at 39 weeks.

Growth

The typical growth pattern for a baby with a ring chromosome syndrome is slow growth in the womb, leading to a low birth weight, followed by failure to thrive and continuing slow growth. In babies and children with ring 13, the pattern of slow growth is especially obvious in those with a small ring and a large deletion. Some individuals with a breakpoint at 13q32 or 13q34 follow a growth pattern that is closer to average.

The range of birth weights is wide, especially in babies with a 13q34 breakpoint and having a mosaic constitution may also influence birth weight. There is little information available on eventual height, and this cannot be predicted from birth weight, but twin girls with ring 13 were around five foot (150 cm) tall as adults, while one *Unique* member with a mosaic make-up is five foot five inches tall (165cm) and another is five foot nine inches tall (175 cm) (Talvik 2000; U).

Breakpoint	Average birth weight	Average weeks at birth
13q11 to 13q22	2lb 15oz (1324g)	37
13q32	5lb 14oz (2668g)	36-37
13q34	5lb 6oz (2440g)	39

Source: *Unique* database

Food and eating

Most babies with ring 13 will need help with feeding as newborns. Their small mouth and jaw, tiny appetite and neurologically-driven difficulties in coping with the complex actions and co-ordination of sucking and swallowing usually mean that breastfeeding is not possible at first. Some babies are able to breastfeed after a spell being fed by nasogastric tube or bottle with an adapted teat for preterm babies, but most babies bottle feed. Typically they feed slowly and because of their small size need feeds every two or three hours at first. Reflux – where milk and stomach contents return readily up the food pipe (oesophagus) – is common and if simple

“ We had problems trying to find a teat small enough for N to feed on as she gagged on most teats. Due to her low birth weight and the small amount she did take, we had to feed her every 3 hours. She did not take to solid food until past 2 years old but can now take solid food normally and can feed herself finger foods but still needs help eating foods like yoghurts – age 3½

measures like careful positioning for feeds and raising the head end of the cot for sleeping do not help, babies may need their feeds thickening or prescribed medication. If reflux is very severe and persistent, it is possible to surgically tighten the valve between the stomach and the food pipe in a procedure known as a fundoplication. A small number of babies are helped by having their feeds and medication given direct into the stomach through a gastrostomy tube. *Unique's* records show that many babies are also late to start solids and their small appetite persists, so they not only eat little but drink little. Constipation is common and with such a small appetite may not resolve with extra fluid and fruit, so that prescribed medicines are needed.

Learning

The great majority of children with a diagnosis of ring 13 will need extra support with their learning. The relationship between the severity of the learning disorder and the amount of chromosome material lost is not straightforward and this makes it difficult to predict the level of eventual ability. Regularly monitoring development is probably the best way to assess the potential outcome in an individual child.

Many people with ring 13 have a level of learning disability that would be described as moderate to severe. This means that academic skills like reading and writing and activities like keeping to time and counting money are always likely to be a challenge. This level of difficulty means that a child diagnosed with ring 13 should, like any other child with a special learning need, be offered extra learning support, both indirectly and directly. Indirect forms of support include occupational therapy (to improve feeding, speech and hand use) and physiotherapy (to improve mobility skills such as sitting and walking). Direct forms of support include portage or other home-based learning programmes and an individually tailored special education programme.

No one knows how severely affected people are whose ring 13 has not been diagnosed, but one young woman, who worked in a daycare centre, could read a newspaper and had no difficulties with speech, was found to have ring 13 with breakpoints at 13p13 and 13q34 only when her daughter was diagnosed with the same ring (Bedoyan 2004).

Many families say that their children are happy and sociable and learn best in a small group setting. Some attend special schools, while others have individual learning support in a mainstream setting. After school, some have gone on to residential college to learn practical skills such as animal care and cookery.

Families say ...

- “ N is mixing well with other children at nursery and is learning to share and take turns. Her strengths are that she is happy and sociable. She does not appear to understand things and then suddenly she will be able to do something – age 3½
- “ A has a wonderful sense of humour and a joyful, loving personality that endears others to him. He refuses to be beaten by his communication problems, has a real love of life and is curious about many things, and likes to know as much as he can about his favourite things. He is now working with numbers to 10 and is quite good at thinking about ideas rather than concrete objects – age 9
- “ K will hopefully go on to residential college for three years. Her strengths are that she is always happy, she loves swimming, animals, listening to others talk, gardening, sleeping and eating. She learns through determination and a sense of humour – age 21
- “ A attends the Welsh Horticultural College for two days a week. His strength is that he lives in the present. He loves the cookery sessions he attends at college, especially eating the product. He has logic and is determined and also has a good sense of humour – age 24
- “ B was able to learn and retain information but never learned to read, and could write only by copying things down. He did learn numbers and could count but has no concept of time. His strengths are determination and stubbornness and, believe me, it has got him through! He did not have a statement of special education need and attended regular schools – age 36

Families say ...

- “C babbles on in her own way and when she tells you something her voice gets higher. She can understand what you say but takes no notice! – age 22
- “F first spoke when she was 2. Today, she finds difficulty in expressing her feelings, and cries a lot when upset – age 20
- “B talks in normal sentences, but her pronunciation can be poor – age 13

Speech and communication

Speech and language delay can be expected in children with ring 13 and in the most mildly affected children may be the presenting symptom. In general, the level of speech difficulty reflects the level of learning difficulty.

Among 17 children and young adults with ring 13 for whom *Unique* has information, the level of delay is very variable. Five young people can carry on a reasonably fluent conversation, although their pronunciation may be hard to understand.

Ten youngsters use some words, but also rely on other communication means including signing, gestures, pictures and communication aids. All of the youngsters with some language have a breakpoint at 13q34. Two people have no words and both have a breakpoint closer to the centromere. In the medical literature, one adult with normal speech and breakpoints at 13p13 and 13q34 has been described (Bedoyan 2004; U).



Mobility and activity

Babies and children with ring 13 show some degree of delay in reaching their mobility milestones. The degree of delay is quite varied and may depend on the breakpoint.

There appear to be two groups in the *Unique* series:

- Babies who rolled around 4 months, sit between 6 and 8 months and start walking between 11 and 21 months
- Babies who were able to sit up, crawl and walk at 3 to 5 years.

Among babies who showed only a slight delay in rolling, sitting and walking, all had a breakpoint at 13q34. At the age of 4, one child had no delay in any mobility skills and was enrolled in classes for tumbling and dancing.

Many babies have abnormal muscle tone – typically their muscles are too floppy (hypotonia). Babies with hypotonia usually need physiotherapy and may need supports to help them to walk. These may be body supports such as standers and walkers until they are able to walk independently and they may also need braces, splints or supportive boots to stabilise particular joints, such as the ankles. Some babies choose to bum shuffle instead of crawling due to low muscle tone in their arms. Once on their feet, children may have a ‘loose’ or ‘sloppy’ gait due to their continuing low tone and flexible joints. This can mean that they are liable to fall easily and some children need extra protection because they do not hold out their hands spontaneously if they fall.

Among the adults with ring 13 in the *Unique* series, mobility does appear to be compromised. Three out of eight adults had shortened tendons in their legs, which needed surgical correction. One adult had unstable knee joints which hampered him from rising from the ground. Hip and ankle joints

could also be stiff and balance remained an issue. While all adults had been able to walk indoors and outdoors at some point, a few relied mostly on a wheelchair outdoors. By his thirties, *Unique*'s oldest member with ring 13 had lost many mobility skills and was entirely dependent on mechanical lifts for mobility (Venugopalan 2001; U).

Fine motor and co-ordination skills

All children in the *Unique* series experienced some level of delay in handling objects, passing them from hand to hand and using both hands together. As well as a general delay in fine motor control and hand-eye co-ordination, some individuals experienced tremor when trying to complete a task and relative hand immobility due to finger contractures. Occupational therapy is helpful and most children were helped by using big grip cutlery and aids for holding pens and markers. The eventual range of manual skill is quite broad, with one adult able to spin, play drums and enjoying craft activities while others have difficulty managing cutlery and writing.



Four years old

Families say ...

- “Generally a very happy, smiling and cheerful child. Occasionally she will have a temper tantrum if she cannot have her own way – age 3*
- “A happy confident little boy, full of fun, great joy to everyone he meets – age 9*
- “He gets violent and aggressive when his routine is changed or something unexpected happens that he does not like. Behaviour has become more of a problem as he has become stronger – age 20*
- “She is normally fine on a day to day basis but can get very moody, tearful and needing reassurance with premenstrual tension. Of late she has become quite deceitful and has got into a habit of spending money on a daily basis. She has become more private as she has got older. She is helpful, placid most of the time, but can be sad and cry a lot – age 20*
- “She is a lovable rogue - never sits still for a long time and still gets up to mischief! She grabs people’s bags or their food in a restaurant! She is very happy, friendly and excitable. Difficult behaviour occurs once a month – age 23*
- “Hyperactive, his behaviour means that he needs constant attention in a structured environment. He gets anxious about going out, and can lash out at times or when his wishes are not understood – age 24*

Behaviour

Information on behaviour comes entirely from *Unique’s* database. There is not enough information to be certain whether there is any particular pattern of behaviour that is typical of ring 13 but it appears more likely that the behaviour shown by most children and adults can be expected from their difficulties with learning and communication and is not specific to their chromosome disorder.

Babies show no remarkable behaviour and if anything are quiet and laid back. As toddlers, some children find their inability to communicate their wishes frustrating and they can be disruptive, showing tantrum behaviour typical of younger children but more intense and lasting for longer. Some families have reported challenging behaviour among children whose wishes are frustrated. A number of families have reported that their children have difficulties in changing routine.

Among adults, some show a level of hyperactive behaviour and are best cared for in a structured environment. Families have also reported agitation, anxiety and occasional bouts of aggression. If this description sounds negative, it should not. Families also report that their children are happy, friendly and helpful.



Right, five years old

Can a child ever live independently?

It is certainly true that some people with ring 13 are able to live an independent life and also true that others lead high quality, fulfilling lives but are not independent. The level of independence that people reach depends chiefly on how much they are affected by their ring 13. Among *Unique*'s membership, all the adults live in group homes or with their own family and need 24-hour support, but this is not of course true of everyone with ring 13. One adult described in the medical literature works in a care home. Within *Unique*, one adult has a personal support worker to ensure round the clock care while his family are out at work. One adult shares a house run by his local mental health trust with four other adults. Some adults achieve a large measure of personal care, but this is not possible for all. Most families, but not all, need support from social services, respite care or nursing services.

Two *Unique* adults have experience of the workplace. One young lady has worked as a catering assistant in a local restaurant and is hoping to gain a vocational qualification that will allow her to work with young children; another has worked in a supermarket, stacking shelves. Social lives have been full and active, including shopping, swimming, listening to music, writing letters, sending texts, treadmill. These aspirations are not possible for all but show that while having ring 13 syndrome, like any disability, may well change lives it does not need to limit them.

Adults out and about





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 Sixto Garcia-Minaur, clinical geneticist, North West Thames Regional Genetics Service and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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