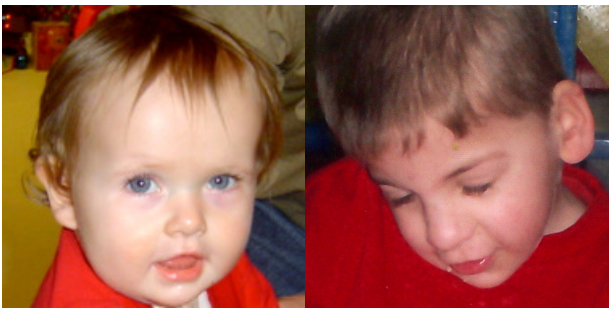
Understanding chromosome disorders

Unique

Supernumerary chromosome 8



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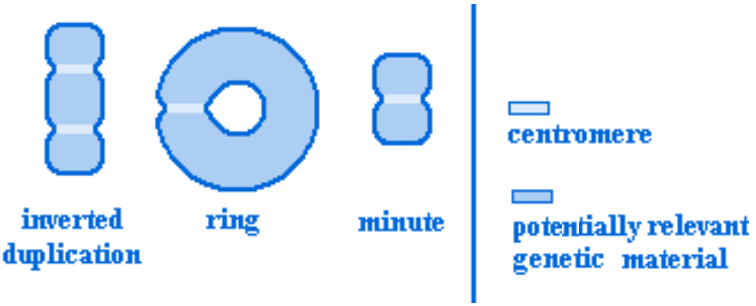
Supernumerary chromosome 8

Supernumerary chromosome 8 means that there is a tiny extra part of a chromosome in all or some of the cells of the body. In addition to the 46 chromosomes that everyone has, people with a supernumerary chromosome 8 have a small extra chromosome made from chromosome 8 material.

The small extra chromosome can have different possible shapes.



It can also have different names. The most common names are:

small supernumerary marker chromosome (sSMC)

supernumerary ring chromosome (SRC), if it's in the form of a ring

Other names you might find in the medical literature include:

small accessory chromosome (SAC)

extra structurally abnormal chromosome (ESAC).

Genes and chromosomes

Our bodies are made up of billions of cells. Most of the cells contain a complete set of tens of thousands of genes which act like a set of instructions, controlling our growth and development and how our bodies work.

Genes are carried on microscopically small, thread-like structures called chromosomes. There are usually 46 chromosomes, 23 inherited from our mother and 23 inherited from our father, so we have two sets of 23 chromosomes in 'pairs'. Apart from two sex chromosomes (two Xs for a girl and an X and a Y for a boy) the chromosomes are numbered 1 to 22, generally from largest to smallest.

Sources & references

The information in this leaflet is drawn partly from published medical research where there are reports of around 40 cases. The first-named author and publication date are given to allow you to look for the abstracts or original articles on the internet in PubMed (at www.ncbi.nlm/nih.gov/pubmed). If you wish, you can obtain abstracts and articles from *Unique*. The leaflet also draws on *Unique*'s database. When this leaflet was written, *Unique* had thirteen members with a supernumerary chromosome 8.

Up-to-date information on extra chromosome 8s is also to be found on the following website: http://www.med.uni-jena.de/fish/sSMC/08.htm. It is rather technical, and if you have difficulty with any of the information there, please contact us at info@rarechromo.org.

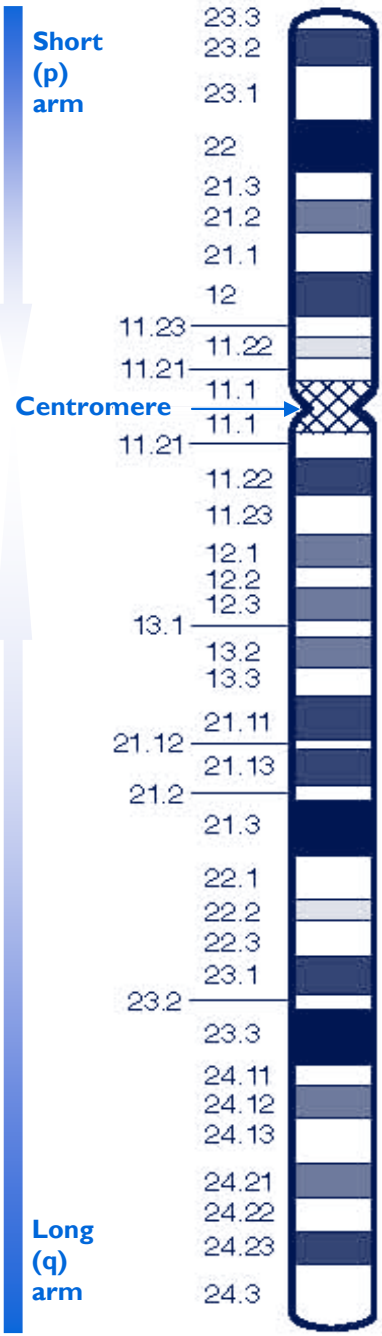
People with an extra chromosome 8 usually have some cells with 46 chromosomes and others with 47 chromosomes (46 plus the extra chromosome). When cells with a different chromosome make-up exist alongside each other, the condition is known as mosaic. People with a small extra chromosome 8 are almost always mosaic. The degree of mosaicism can be different in every body tissue. So it can be different in the blood, the skin, the lungs, heart, brain and so on.

Chromosome 8 is a medium-sized chromosome and contains 700-1,100 genes out of the total of 20,000 to 25,000 genes in the human genome. The extra chromosome usually only contains a few of these genes but it's generally believed that these extra genes cause any clinical difficulties that someone with a supernumerary chromosome 8 may face.

You can't see chromosomes with the naked eye, but if you stain them and magnify them under a microscope, you can see that each one has a distinctive pattern of light and dark bands. The diagram on this page shows the bands of a normal chromosome 8. These bands are numbered outwards starting from the point where the short and long arms meet (the centromere). A number starting with a 'p' is in the short - petit - arm (at the top in the diagram) and a number starting with 'q' is in the long arm (at the bottom).

Some chromosome bands contain no important genes and only irrelevant genetic material (called heterochromatin). Having extra material from bands containing heterochromatin does not generally affect development. Other bands contain relevant genetic material (called euchromatin). Having extra euchromatin is more likely to affect development but does not always do so.

Some people with an extra chromosome 8 have a single extra copy of the genes in the euchromatin. Other people have two extra copies. People with an inverted duplication (*see image on page 2*) have two extra copies.



Chromosome 8

Your child's karyotype or molecular results

Your geneticist or genetic counsellor will tell you more about what chromosome material your child's extra chromosome consists of. You will almost certainly be given a karyotype, a shorthand code for the image of your child's chromosomes. This may show the points where the chromosome has broken. A karyotype for a boy with supernumerary chromosome 8 might look like this:

mos 47,XY,+r(8)(p12q21.3)[28]/46,XY[22]

mos	= mosaic or cells with a different chromosome make-up exist alongside each other
47	= total number of cells in the first cell line is 47. There is one extra chromosome
XY	= the two sex chromosomes, XY for males; XX for females
+r(8)	= the extra chromosome consists of material from chromosome 8 and is in the form of a ring
(p12q21.3)	= the chromosome has broken in the short arm at band p12 and in the long arm at band q21.3, so the extra chromosome consists of the material between these two points, including the centromere
/	= information after this sign is about a different cell line
46,XY	= the chromosomes in the second cell line are those of a normal male
[28] [22]	= these figures tell you that 50 cells were analysed, of which 28 contained the extra chromosome and 22 normal chromosomes.

Comparing your child's karyotype with others, both from the medical literature and within *Unique*, can help to build up a general picture of what to expect. But there will still be differences, sometimes quite marked, between your child and others with apparently similar small extra chromosomes. It is very important to see your child as an individual and not to make direct comparisons with others with the same karyotype.

Diagnosis

The extra chromosome cannot usually be identified accurately under a microscope. It may be so tiny that it is even hard to know which chromosome it comes from. The origin and amount of extra material can usually only be identified using new, sensitive molecular techniques such as FISH (using specific DNA probes that show up in fluorescent colour) and array-CGH (also known as microarrays), a way of analysing thousands of different DNA sequences at the same time. In terms of predicting the outcome for any individual child or pregnancy, it is important to be as precise as possible about the breakpoint in each arm of the chromosome.

Does the proportion of cells with the extra chromosome matter?

This question refers to the numbers in square brackets in the karyotype. It tells you more about the mosaicism – the cell line with the extra chromosome and the cell line with normal chromosomes. Although you will often be given this information, in fact it probably doesn't help in understanding the effects. The cells tested are usually only from blood or sometimes from scrapings inside the cheek - but this doesn't tell you about the proportion of cells with the extra chromosome in other tissues of the body, such as the heart, or liver, or lungs or brain.

Can someone with a supernumerary chromosome 8 have similarly affected children?

In adults, especially men, with a small extra chromosome 8, fertility can be affected but some people will be able to have children. The children can inherit the extra chromosome. The chance of inheriting a small supernumerary chromosome from one's mother is twice as high as from one's father (Liehr 2006).

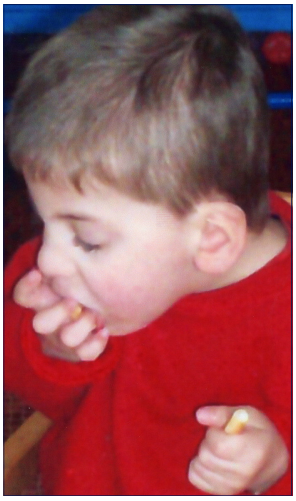
A small extra chromosome containing no genetically relevant material can run in some families from generation to generation without causing any problems. But when a parent passes on a small extra chromosome that contains genetically relevant material, the baby can have more or fewer affected cells than the parent - and this of course can change its effects (Rothenmund 1997).

How will the extra chromosome affect my child?

The effects of the extra chromosome depend mostly on whether it contains genetically relevant or irrelevant material, how much material it contains and from which bands of chromosome 8. Your geneticist or genetic counsellor will be able to tell you whether the extra chromosome contains genetically relevant material or not and which part of the chromosome it comes from.

To discover about the possible effects, one good way is to contact info@rarechromo.org and ask for help in selecting relevant cases from the website at http://www.med.uni-jena.de/fish/sSMC/08.htm. You can then take a list of these cases to your genetic specialist. You can also look in this leaflet for the relevant page/s (6-15). The information in these pages was correct when we wrote this leaflet in early 2009, but the picture can change as new babies and children are diagnosed and reported in the medical literature.

15 months

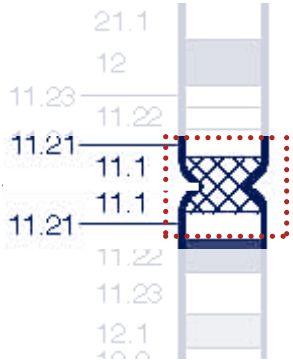


Four years

Are there people with a supernumerary chromosome 8 who are healthy and whose development has not been affected?

Yes, there are. When the extra chromosome only contains genetically irrelevant material, development isn't affected. And there are one or two children with an extra chromosome made up of material from very close to the tip of the short arm (8p) who appear to be developing quite normally (Herry 2004; de Pater 2005). There are around 15 other reports in the medical literature of people with normal development where the size of the extra chromosome 8 hasn't been established. These include: a 31-year-old woman with apparently completely normal development, a normally developing seven-year-old girl and a normally developing two-year-old (Gravholt 1995; Tonk 2000; Daniel 2003).

Extra chromosomes containing only genetically irrelevant material from around the centromere between 8p11.21 and 8q11.21

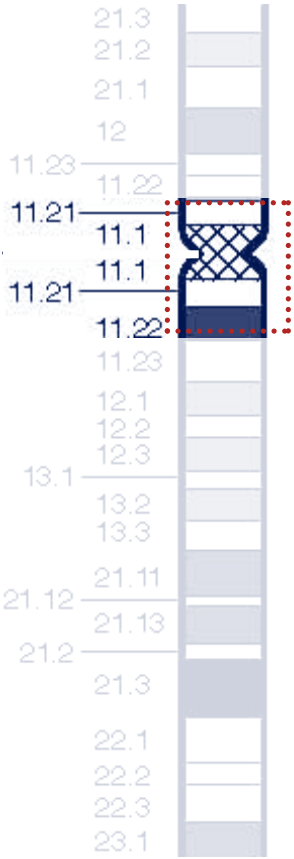


Chromosomes that contain no genetically relevant material are probably harmless and do not affect development.

The chief effect of these otherwise harmless extra chromosomes is that they may affect fertility.

Chromosomes containing only genetically irrelevant material do not generally extend beyond band 8p11.21 in the short arm or beyond band 8q11.21 in the long arm (Starke 1999; Gole 2005).

Extra chromosomes consisting of small amounts of material including the centromere and as far as 8p11.21~22 in the short arm or 8q11.22 in the long arm



Only a small number of cases have been characterised in detail. From these, the most likely features in the affected child appear to be:

- Some degree of developmental delay
- Delay in acquiring language
- Some difficulties with social skills
- Some behaviour difficulties including problems with attention and activity levels or autistic-like behaviour

(Daniel 2003; Loeffler 2003; Liehr 2006; Bettio 2008; *Unique*).

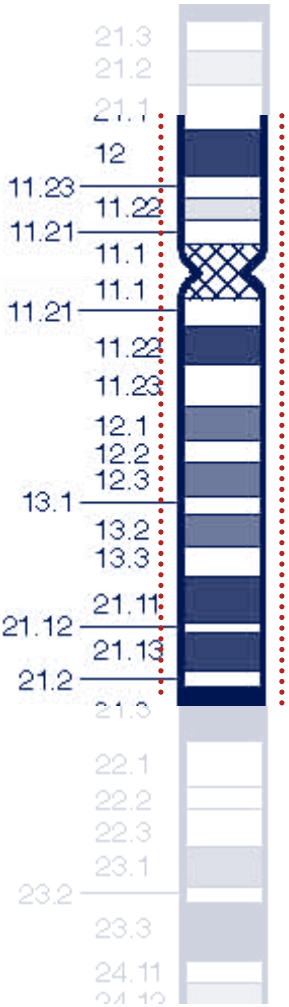
Additional details from a description of a *Unique* member at 15 years: normal birth weight; early feeding difficulties and breast refusal; no underlying health problems or physical abnormalities; motor development just within normal limits and normal mobility – sitting, walking, running etc; late acquisition of fine motor skills but no limitation; need for specific support with learning. Particular difficulties with mathematics and abstract subjects including history and geography. Attended mainstream (regular) primary school with a 1:1 teaching aide. At 15 years, attends a special education school where the curriculum is styled to his level and he is taught in small groups without an aide. He reads comics, teen magazines, TV guides, the weather and movie sections of the newspaper. His expressive language is delayed, so he can only express himself in a limited way and

it is not always clear how much he understands. He spoke his first words at two and by five a few more words, but at 6-7 had a great leap in vocabulary. At 15, he speaks in somewhat simplified and repetitive sentences.

He has had a diagnosis of atypical autism (pervasive developmental disorder) since he was five. He also has some obsessive-compulsive behaviour. He responded immediately to Applied Behavioral Analysis therapy (ABA) at 5-7 years and made great strides in all major skill areas. At 15, he is shy with children of his own age, doesn't always understand what they say or mean and is worried he will act inappropriately. As a child between 1.5-7 years he had big sleep problems, waking for hours every night. A gluten/casein-free diet helped his sleep, as did melatonin before bedtime.

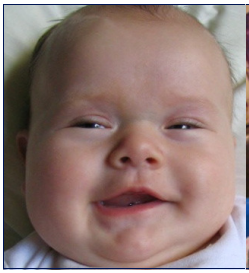
Extra chromosomes containing material from beyond band 8p11.22 in the short arm and band 8q11.22 in the long arm

Three young children within *Unique* each have a small marker chromosome made up of material from the short and the long arms of chromosome 8. In one child the breakpoints are assessed to be at 8p11.21 and 8q11.21; in the second, with a ring, at 8p12 and 8q11.21 and in the third child with a slightly larger ring they are at 8p21 and 8q21.3 (*Unique*).



The first child, the second of two children, was born at full term after a healthy pregnancy. She was born with two extra nipples, but these caused no initial concern. As a baby she was very congested and often brought breast milk back up after feeding, but no related allergies were found. At 2 years, she was diagnosed with asthma and enlarged adenoids; the salbutamol/fluticasone and steroid adenoid sprays have helped a lot to relieve congestion. She has chronic nasal and upper respiratory congestion and has glue ear, causing a form of temporary hearing loss, which is treated with tubes. Her growth was initially at the 95th percentile for weight and height, but at almost 4 years is now at the 25th percentile and she is slightly small for her age. A thyroid check at the age of 2 came back normal. Her balance and stability improved after the tubes were inserted. She has developmental delay and a low muscle tone, and is happy, active and sociable. Further, she has responded very well to occupational and physical therapy. At 2½, she was able to do things overall that an average 17-month-old might do and by the age of 3, she was walking stably and trying to jump, kick and catch a ball. She was able to drink and finger-feed herself alone and she has started to use a spoon and fork. By 3½, she could wash her hands alone, count to three, identify opposites, walk upstairs and dress with help, but was not yet toilet trained. She

enjoyed playing with toy babies and Barbie dolls, dancing, swimming, playing hide and seek, wrestling and playing superheroes as well as looking at books, watching TV and singing. She has a lot of ingenuity and compensated for her lack of language with facial expressions and gestures, and very strong social skills. She was still using 2-3 word phrases and had difficulty making *p* and *f* sounds and saying the ends of words, although recently (3 years, 10 months) she has not progressed in her language ability. She was enrolled in an early education programme where she showed that she was extremely determined and resourceful and learned best with 1:1 teaching and repetition.

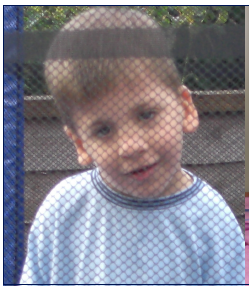


3 months **14 months** **3 years**

❝ An absolute joy! She is exceptionally happy and outgoing, will even introduce us to people and ensures all children are included in play.

The child with the somewhat larger ring was born slightly early at 37 weeks but at 3.2kg (7lb 1oz) was a healthy weight. The main complication in pregnancy was very marked excess amniotic fluid. He spent the first two weeks of his life in special care and was tube fed, partly because he had difficulty coordinating breathing with feeding. He also tended to bring milk back during and after feeds (reflux) and this persisted until he stopped milk feeds. As a newborn baby his most obvious feature - apart from some subtly different facial features and a sacral dimple at the base of the spine - was talipes (club foot), requiring casting to realign the feet ready for walking. This has been completely successful and having started to walk at 3, he is now trying to run. He is generally healthy but has had repeated respiratory infections as a young child. The only regular medicine he takes is slow-release melatonin to help him stay asleep.

Developmentally, he is starting to toilet train at four years old, he can do basic signing, can say Hi, can count to 10 and attends a special needs school four days a week and a mainstream primary with his mother one day. The hope is that he will eventually have a 50:50 dual placement. He eats independently and drinks from a standard beaker. He loves CBeebies, singing, mobile phones, the touch screen computer, the family dog and jumping on his large trampoline with his other siblings!



3 to 4 years

❝ Above all, we have tried to treat him like other children.

Isochromosome 8p: mosaic tetrasomy 8p

People with this rare chromosome disorder have a small extra chromosome - called an isochromosome - in some cells of the body. The isochromosome consists of two copies of most or all of the short (p) arm of chromosome 8. In these cells there are four copies of the short arm of chromosome 8 instead of the usual number of two. Geneticists call a condition where there are four copies of part of a chromosome a tetrasomy. Having an isochromosome 8p causes a disorder known as mosaic tetrasomy 8p. The effects of this disorder are caused by the genes on the extra chromosome.

Mosaic tetrasomy 8p is very rare: since the first case was reported in 1988, only a total of 17 cases have been reported in the medical literature. *Unique* has three members with this disorder.

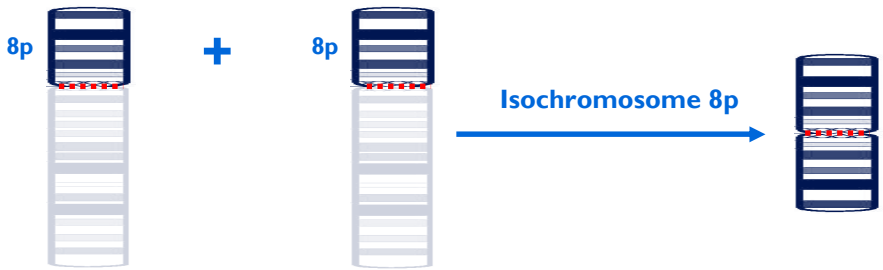
When there are very few cases, with reports only of babies or young children, it's not possible to be certain about the full range of effects of a particular chromosome disorder. In particular, in a mosaic form of a disorder, effects can range from very minor to extremely severe.

But some possible effects of mosaic tetrasomy 8p have been seen repeatedly. Some of these features are also seen in people with mosaic trisomy 8 (where some cells contain a single copy of the whole of chromosome 8).

These features are:

- A degree of developmental delay. The typical range is from moderate to severe. Speech and language appear to be specifically affected
- Structural anomalies of the brain. These typically include agenesis of the corpus callosum (absence of the band of nerve fibres that links the two hemispheres of the brain). They also include enlargement of the fluid-filled spaces within the brain
- Heart problems. These may be simple, such as a hole between the two upper or lower heart chambers, or more complex, involving a number of defects. Children may outgrow some small defects and others are correctable by surgery but this may not be possible for all
- Fused or oddly formed bones in the spine (vertebrae); extra or missing ribs
- Spinal curvature

(Kristoffersson 1988; Robinow 1989; Roskes 1990; Fisher 1993; Newton 1993; Tilstra 1993; Digilio 1994; Schrander-Stumpel 1994; James1995; Winters 1995; Napoleone 1997; Le Bris 2003; López-Pajares 2003; Nucaro 2006; *Unique*)



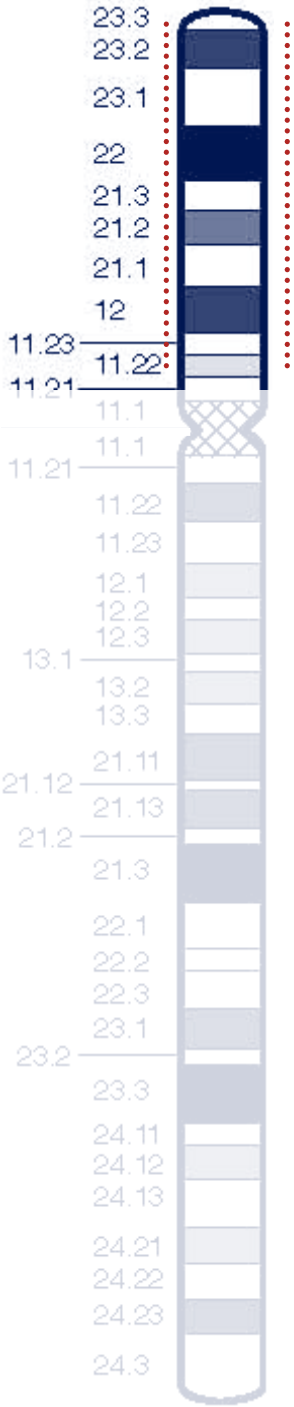
Chromosomes containing only genetically active material from the short arm beyond band 8p11.22

Of the eleven cases reported in the medical literature, each almost certainly has a different precise breakpoint in the short arm. Many of the reports are also quite incomplete and this limits what can be said about the general effects of an extra chromosome consisting of material from the short arm of chromosome 8 (Ohashi 1994; Batanian 2000; Anderlid 2001; Voullaire 2001; Daniel 2003; Demori 2004; Herry 2004; De Pater 2005).

The **development** of some children with a small extra chromosome between the tip of the short arm and band 8p22 was not affected or only affected in a very slight way. One baby with a chromosome made up of two copies of material from the tip of the short arm to band 8p22 was developmentally ahead of her chromosomally normal brother. A baby with a smaller extra chromosome to band 8p23 was developing normally at two years (Herry 2004; De Pater 2005).

But others with similar extra chromosomes were developmentally affected. A 21-year-old with a ring consisting of most of the short arm of chromosome 8 was judged to have some learning difficulties, with an IQ of 80-85 (Daniel 2003). One 8-year-old child with an extra chromosome as far as 8p23.1 was developmentally delayed but 'good at sports'; a 13-year-old with an extra chromosome as far as 8p23.11 had below average learning abilities and an IQ of 75 (Voullaire 2001). And a 2-year-old with an extra chromosome as far as 8p23.1 had a developmental quotient of 73.

Where the extra chromosome consisted of material from the centre of the short arm between 8p10 and 8p23.1, one child was sitting and moving just within normal developmental limits and at three years was assessed developmentally to be at a 25-month level. In terms of speech, he was a slow starter but putting words together at 3 years (Demori 2004). A child with an extra chromosome made up of material



from between the centromere and bands 8p12 was developmentally normal in his pre-school year but lagged a year behind academically by age 5 (Batanian 2000). A two-year-old with an extra chromosome as far as 8p10 was 10 months delayed at the age of two and had a mild learning disability at 8 (Anderlid 2001).

In terms of **behaviour** and **social ability**, one boy showed autistic-like behaviour at three years of age, with stereotyped conduct, avoiding gaze and repetitive language sounds (Demori 2004). Another boy had unspecified behaviour problems at the age of three (Anderlid 2001), a third was treated for attention deficit hyperactivity disorder at the age of nine and had continuing behaviour problems at 13 (Voullaire 2001) while a fourth was treated for attention deficit disorder with methylphenidate (Ritalin) and imipramine when he was five. He was also on anti-epileptic medication for seizures (Batanian 2000).

As for **growth** and stature, this is generally not commented on but one adult woman was 159cm (5' 3") tall, a 2-year-old girl was slightly short for her age and a 13-year-old boy was in the shortest three per cent of the population for his age (Ohashi 1994; Voullaire 2001; Daniel 2003). Features that might be noticed at birth were unusual although some babies had subtly atypical facial features and one baby had several large birthmarks on her back (Batanian 2000).

One boy was also born with a very small penis, **undescended testicles** and **hernias** in the groin. Treatment for undescended testicles is usually needed if the testicles do not descend naturally in time. The testicles can be brought down in a short operation under general anaesthetic called an orchidopexy. Inguinal hernias also usually need surgical repair (Voullaire 2001).

Six of the children appear to have enjoyed generally good health while four had significant illness.

Two boys had repeated respiratory infections in the first year of life, in one case as well as asthma and a milk allergy. Both had a significant **heart condition**, persistent ductus arteriosus (PDA), in which a channel taking blood to the lungs during fetal life fails to close as usual shortly after birth. In both cases the channel was surgically closed in the second year of life (Ohashi 1994; Voullaire 2001). A third baby had a heart defect known as total anomalous pulmonary venous return, in which the vessels that bring oxygen-rich blood back to the heart from the lungs are incorrectly connected. Treatment depends on the baby's condition but can include medication to help the heart and lungs work more efficiently and surgery to reconnect the blood vessels. At the age of five, this child developed thrombocytopaenia, where the number of platelets in the blood is reduced, leading to bleeding into the skin, spontaneous bruising and lengthy bleeding (Batanian 2000).

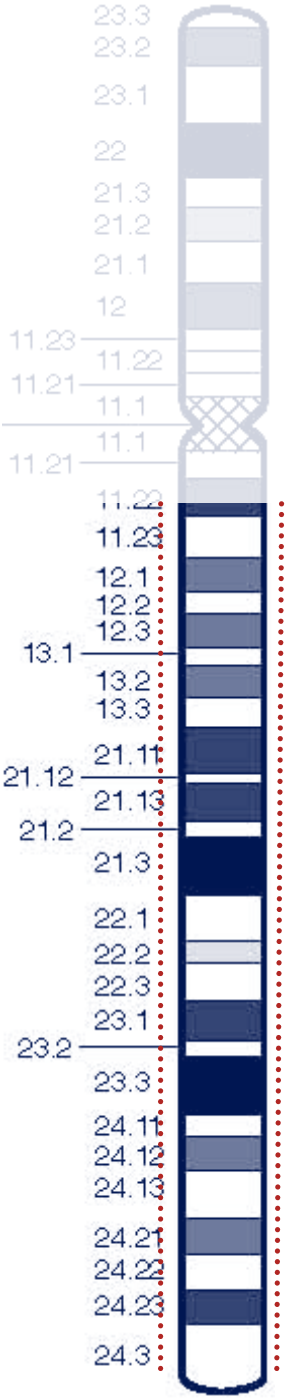
As for other conditions, one baby was noted during pregnancy to have enlarged fluid-filled ventricles within the brain and this was confirmed as hydrocephalus at birth (Batanian 2000). **Puberty** was precocious in one girl, starting at the early age of seven, while a boy in whom puberty was delayed required hormonal stimulus (Batanian 2000; Voullaire 2001).

Extra chromosomes containing genetically active material from the long arm distal from 8q11.22

At least seven cases of rings and markers are listed on http://www.med.uni-jena.de/fish/sSMC/08.htm. Here two cases are described from the medical literature. The extra chromosome material is quite different in each case. So it isn't possible to draw any general conclusions.

An 8-year-old girl with a ring chromosome made up of material from the centromere to band 8q21.1 had experienced some developmental delay, sitting and walking quite late, although this was partly explained by repeated operations to correct her club feet. Her fine motor skills – hand use and coordination – were on track and intelligence-wise, she was about a year behind her peers at school. All the same, she was much stronger in some aspects of learning than in others. Her chief difficulties were social and communicating, not least because she had hearing loss and used sign language. Physically, she was healthy. She had narrow shoulders and subtly unusual facial features that were less obvious than when she was a baby (Anderlid 2001).

A 16-year-old girl with an extra chromosome made up of two copies of the material from the tip of the long arm to band 8q23.3 was born prematurely at 32 weeks and initially tube-fed. As a newborn baby she showed one or two unusual features, including bent fifth fingers, a curved sole on the feet and increased muscle tension (hypertonia), reducing the ability of muscle to stretch. Her fists were clenched and she couldn't fully open her elbows, knees or hips. This feature remained so that as a teenager she couldn't fully open her fingers. She remained in the smallest five per cent of the population for her age. In terms of development, she showed global delay, walking at 18 months and with an IQ of 45-50, suggesting moderate learning disabilities. When she was almost 11, she knew her colours and the alphabet. By 15, she could count to 27 and could undress, but couldn't read and needed help with dressing. Behaviour-wise, she was sociable and affectionate but teachers described her as hard to handle and having only a short attention span. Her increased muscle tone remained and she walked on her toes. Subsequently her ovaries were found to be abnormal (they contained multiple cysts) and she grew some facial hair. She later developed the autoimmune disorder myasthenia gravis, a nerve condition that leaves the muscles around the body weakened (Reddy 2000).



Extra chromosomes of unknown extent

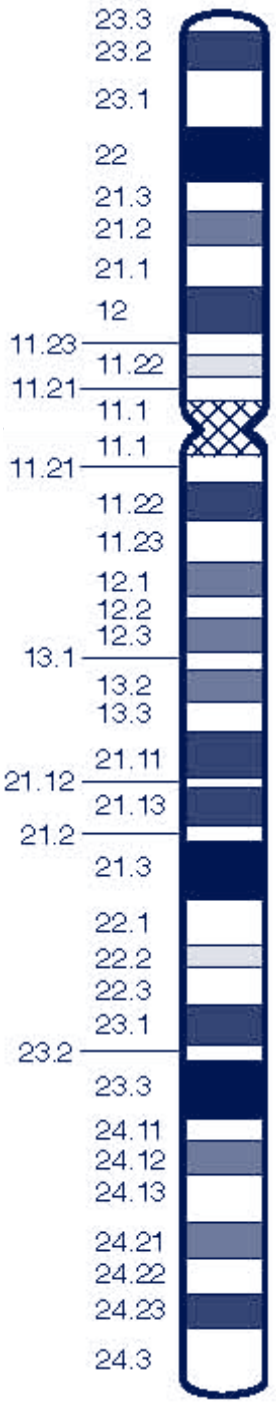
Seventeen people have been reported in the medical literature with an extra chromosome, frequently a ring chromosome, made up of material from chromosome 8 but with no specification of break points, leaving it uncertain what material it contains. Two of them, including a 31-year-old woman, have apparently entirely normal development and were discovered by chance or during routine screening. The comments below do not relate to these two (Plattner 1993; Daniel 1994; Butler 1995; Gravholt 1995; Hastings 1999; Tonk 2000; Daniel 2003; Yilmaz 2005). Nine *Unique* members aged from 6 to 37 years have a similar extra chromosome 8, again usually a ring chromosome.

In terms of **development**, all have shown some delay but the extent is very variable. Some babies have low muscle tone and are late to acquire head control and to roll over. Babies learned to sit alone between seven and 16 months and to walk between 14 months and four years, in some cases initially using a walking frame. Three babies were born with talipes (club foot) requiring casting and sometimes surgery to acquire a normal walking position and in these children walking was understandably delayed. Others were born with more minor effects, such as mild clawing of the toes. Once on their feet, some children have remained unbalanced and prone to tumbles while others move around entirely normally. One adult of 27 years has developed arthritis-like pain in his knees while moving around.

As far as **fine motor skills** are concerned, some are unaffected, while unbending fingers and limited joint movements in the hands and arms make for dexterity problems for others. **Speech** is generally delayed but understanding is usually more competent than expression. Four of the nine *Unique* children had a temporary or permanent hearing impairment with obvious impact on their speech development. Despite this, some children are communicating with words as well as gestures, vocal noises, facial expressions and objects of reference by their third year, while speech emerges in others somewhat later.

❝ He communicates with signing, pushing, pulling, gestures and vocal noises and now says Mum and Dad - *6 years*

❝ At the age of three he would say very few sentences composed of two words. He still sometimes has a problem expressing himself and maintaining a conversation. He understands well, although on occasion it is impossible to make certain situations understood - *27 years*



Children do typically need **learning** support at school, with their abilities ranging from mildly to severely impaired, with many in the moderate range of ability. Typically, children will require a statement of special educational need and will either attend a mainstream school with support or a special school better adapted to their particular needs. Some children learn to read and write but this is not possible for all. Many children have difficulties with concentration and attention as well as a communication deficit that is reminiscent of autism and undermines their ability to learn efficiently until they are correctly diagnosed and appropriately managed.

❝ He enjoys music and drawing, is good at puzzles, and loves to take things like old radios apart. He likes to fix things and at times is helpful which makes him very happy. He is very slow but likes to do things well even if it takes all day or more - *27 years*

In terms of **behaviour**, children are generally described as well behaved but prone to frustration (angry, impatient, cross, challenging) when they can't communicate what they want or get their own way. Autistic tendencies may undermine social skills but this is not universal and some youngsters are easy-going and well-liked.

❝ He can be very calm and well-behaved but also very demanding with tantrums if he doesn't get his own way. He likes to be with other children and to be cuddled. He's strong-willed: when he wants something he doesn't give up until he gets it - *6 years*

❝ Most of the time he is a very peaceful person but there are days where he gets angry easily but this has never been a problem. He's a little shy socialising in the beginning but when he feels comfortable he enjoys the company of others. He is well liked by everyone. He's very special with us, his parents, when we are sick. He worries a lot and takes good care of us. He loves to buy me things and knows just what I like because he goes everywhere with me - *27 years*

In terms of **growth**, information is available on six people, only one of whom was short for his age. All others were of normal height or unusually tall with a slender build. **Facially** the one feature of note is large ears which in one child were corrected with plastic surgery to give them a more normal appearance.

There is little information on **feeding** and it seems that some babies at least will breast feed without particular difficulties and move on to thicker feeds and solid foods at an appropriate age. Others will need support with feeding and may thrive better when fed from a bottle. Reflux, where feeds return up the food passage and may be vomited, has occurred in some children and will need careful management with appropriate seating positions and possibly thickened feeds and prescribed medication to help prevent feeds from returning up the food passage. Children may be late to wean on to solid foods and need them puréed or finely chopped for much longer than other children.

In terms of **general health**, one 15-year-old developed **seizures**, but they responded to medication, which was then stopped after two years. One child was diagnosed with leukaemia. One baby was born with a hole between the two lower (pumping) chambers of the **heart** and this was successfully repaired surgically. Four children had some degree of kidney involvement: one had a small, non-functioning **kidney** removed; another had an arrangement known as horseshoe kidney, where the bottom points of the two usually separate kidneys are joined, creating a U (horseshoe) shape; another child had the ureters that lead from the kidneys to the bladder surgically reinserted;

another young child with a swollen kidney (hydronephrosis) took low-dose antibiotics to protect his kidneys. Minor **genital** anomalies are not uncommon in children with a rare chromosome disorder and this was true of four children with a small extra chromosome 8: one boy was born with hypospadias, where the hole normally at the end of the penis is on the underside instead; another had unusually small testicles, with one not fully descended into the scrotum; another had a very small opening for urine, corrected surgically; and one girl was born without a clitoris. A developing spinal curve is a feature of the condition known as trisomy 8 mosaicism and is seen in people with an extra part of chromosome 8: the curve may be sideways or forwards and can be slight, needing no more than monitoring or more severe, needing bracing or even surgery.

Eyesight is affected in six of the *Unique* members, although problems are varied, including glaucoma (a rise in the internal pressure within the eye), a squint (strabismus), short sight and Duane's syndrome (restricted turn of the eye). Three members have a degree of loss of vision but this is not so severe in any as to need special teaching.

How did the extra chromosome come about?

Some small supernumerary chromosomes are inherited. The mother or the father has the same small chromosome. A blood test to check the parents' chromosomes will usually show if this is the case.

But most small extra chromosomes occur in children both of whose parents have normal chromosomes. Geneticists call this de novo (dn), meaning that this has occurred as a new event. These extra chromosomes are caused by a change that occurred when the parents' sperm or egg cells were formed or around the time of conception.

There is still quite a lot of uncertainty about the events that caused the small extra chromosome to form. But what is known is that as a parent you cannot change or control them. Children from all parts of the world and from all types of background have small extra chromosomes. No environmental, dietary or lifestyle factors are known to cause them. There is nothing that either parent did before or during pregnancy that can be shown to have caused the extra chromosome to form, and equally nothing could have been done to prevent it.

Can it happen in another pregnancy?

When the parents' chromosomes are examined they are usually found to be normal. Where both parents have normal chromosomes, it is very unlikely that a second child will be born with a supernumerary chromosome 8.

When, as occasionally happens, one parent has a supernumerary chromosome 8 themselves, the chance of passing it on is theoretically as high as 50:50 in each pregnancy, but in reality is somewhat lower.

Support and Information

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Join Unique for family links, information and support.
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