

# Ring 18



## Sources

The information in this leaflet comes from the medical literature and a survey of family members of

*Unique*. *Unique* is very grateful to the families who took part in the survey.

The medical literature largely contains descriptions of people with Ring 18 written by doctors to help other doctors make accurate diagnoses. Some of the features they mention, such as odd-shaped ears, are not very important to parents or to their child. Very obvious or extreme problems such as club feet or heart conditions are more likely to be mentioned and this can create a picture that is unnecessarily gloomy. No one knows how many people there are with Ring 18 who never come to doctors' attention.

## Ring 18

Ring 18 Syndrome is a rare genetic condition caused by having an unusual chromosome.

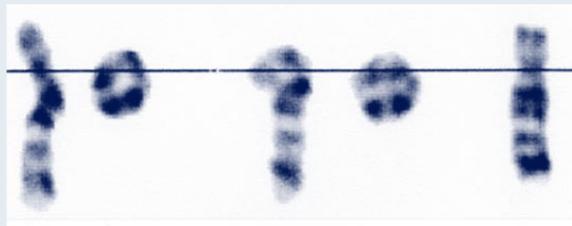
### What is a chromosome?

The human body is made up of cells. Inside most cells is a nucleus where genetic information is stored in genes which are packaged into chromosomes.

Chromosomes are large enough to be studied under a microscope and come in different sizes, each with a top (short) arm called p and a bottom (long) arm called q. They are numbered from largest to smallest according to their size, from number 1 to number 22, in addition to the sex chromosomes, X and Y. A normal, healthy cell in the body has 46 chromosomes, 23 from the mother and 23 from the father, including one chromosome 18 from each parent.

In people with Ring 18, the ends of one chromosome 18 have joined up to form a circle. When this happens, genes are usually lost from either the p arm or the q arm or both, and it is presumed that these missing genes cause most of the signs of Ring 18 Syndrome.

Most people with Ring 18 have the ring chromosome in all their body cells. Some people have some cells with the ring chromosome and some with normal cells. This is called mosaicism and your geneticist will tell you if this is what your child has. The effects of the ring are less easy to predict in someone with mosaicism but they can be milder (Fryns 1992; Stankiewicz 2001).



Ring chromosome 18

## Main features

People with Ring 18 differ a lot from each other and while some people appear completely unremarkable, others are more severely affected. You will find here lists of 'typical' features. Most people have features of 18q- (having lost material from the end of the long arm), some have features of 18p- (having lost material from the short arm) and some have a mixture. Your child will not have all of the typical features or even most of them but he or she will almost certainly have some (Stankiewicz 2001).

### Features more common in people who have lost the end of 18q (18q-)

- Very narrow or closed ear canals, usually with hearing loss. Ears may be abnormally formed, with a prominent antihelix (see below, right).
- Unusual feet: including features such as rocker bottom feet (with a convex sole, without an instep or arch), abnormal or overriding 2nd and 3rd toes, talipes (club foot).
- Congenital heart defect.
- Unusual hands: including features such as tapering fingers, low set thumbs, incurving little (5<sup>th</sup>) fingers.
- In boys, minor genital abnormalities, such as the hole at the end of the penis being on the underside (hypospadias).
- Increased chance of developing juvenile rheumatoid arthritis. This causes inflamed and stiff joints.

### Features more common in people who have lost the end of 18p (18p-)

- Difficulty resisting infections.
- Holoprosencephaly. This is a developmental defect that occurs when the brain and face are forming. The forebrain does not grow forward and divide into two hemispheres as it is supposed to do. The severity varies, with the least affected people having some fused structures and the most severely affected having a single brain mass instead of two separate lobes. Most people come between these two extremes. The middle of the face is often affected as well: one of the mildest effects can be the growth of a single front tooth; one of the most severe, a single eye.

## References

The text contains references to articles published in the medical press. The first-named author and publication date are given to allow you to search for the abstracts or original articles on the internet in PubMed. If you wish, you can obtain abstracts and key articles from *Unique*. Information from the *Unique* survey is marked (U).

## Supernumerary Ring 18

There is an extremely rare type of Ring 18 that occurs when a third, extra chromosome 18 is present as a ring. This is called supernumerary Ring 18 and the features are different to those of the Ring 18 disorder described in this leaflet.



## How common is Ring 18?

Ring 18 is rare but no-one can be certain how uncommon it is because a few people have the disorder without obvious symptoms.

Around 70 people have been described in the medical literature and the Chromosome 18 Registry knows of more than 120 people.

Out of 82,000 people whose chromosomes were investigated at a major European genetics centre, ten had a Ring 18 (Fryns 1992; Schinzel 2001).

- Small jaw (micrognathia).
- Tooth decay.
- Ptosis (drooping of upper eyelids).

## Features common in people who have lost either end of chromosome 18

- Delayed development.
- Some degree of learning difficulty.
- Floppy muscles (low muscle tone, hypotonia).
- Failure to thrive (difficulty putting on weight and growing).
- Short stature with a shortage of growth hormone.
- Small head and brain (microcephaly).
- Skin folds across the inner corners of the eyes (epicanthal folds).
- Abnormalities of the roof of the mouth (palate).

## Other features

Many other unusual features have been described in people who have lost material from the ends of chromosome 18. However, not enough people have been described yet to be certain that these features are caused by loss from chromosome 18 or Ring 18 Syndrome.

## Experiences of pregnancy

A small group of 12 families have told us about their pregnancy experiences. They are varied but the largest number of mothers carrying babies with Ring 18 experienced no pregnancy problems and only discovered that their baby was affected after the birth. Four babies were small for dates and two mothers had a low level of amniotic fluid. One mother had excess amniotic fluid and two babies were induced two to three weeks after their due date. Other experiences may have no link with Ring 18: one mother had a high cholesterol level while pregnant, one mother was carrying twins but lost one, one mother had pancreatitis (inflammation of the pancreas) during pregnancy and had her gallbladder removed (U).

## Newborn experiences

Out of 10 families who told us about their baby's experiences as a newborn, seven had feeding difficulties,

with a poor sucking reflex and failure to put on weight. Two babies developed neonatal jaundice and two others were very floppy. Two babies developed severe infections while still in hospital, in one case affecting the stump of the umbilical cord, followed by an RSV (respiratory syncytial virus) infection (U).

## First signs

Of 11 families who told us what the first signs were that anything was wrong with their baby, four were healthy and looked quite normal at birth but were slow to reach their developmental milestones. Five other babies had obvious signs at birth – one was very small for dates, two had a cleft palate and two had club feet (talipes). One baby was diagnosed after she developed seizures at four months and one after a severe infection. All babies were diagnosed before their first birthday (U).

## Diagnosis

Ring 18 is diagnosed by examining the chromosomes in white blood cells. The ring chromosome is obvious under a microscope but molecular genetic tests are needed to discover more precisely how much material and which genes are missing. Laboratories offer different molecular tests including FISH ( a technique called chromosome painting) using whole and partial chromosome 18 paints (Stankiewicz 2001).

## Appearance

Most children – typically four children out of five - are very short compared with other family members. If they do not have other obvious problems such as a cleft lip, ptosis (a drooping upper eyelid) or talipes, they may look little different to other children and even if they do, after surgery there is usually little, if anything, to see of the original problem. Even problems that some think of as cosmetic can be corrected surgically. Some, such as the fine, curly and fair hair that some children with Ring 18 have, can just be admired.

## Growth

People with Ring 18 are generally short as children and become short adults. Without treatment, predicted adult height is around five foot (150cm).

## Families say ...

“ Tatum has been on growth hormone since she was 11 months old and has done wonderfully. She caught up in height and weight at first and it also helped her gross motor skills - age 3.

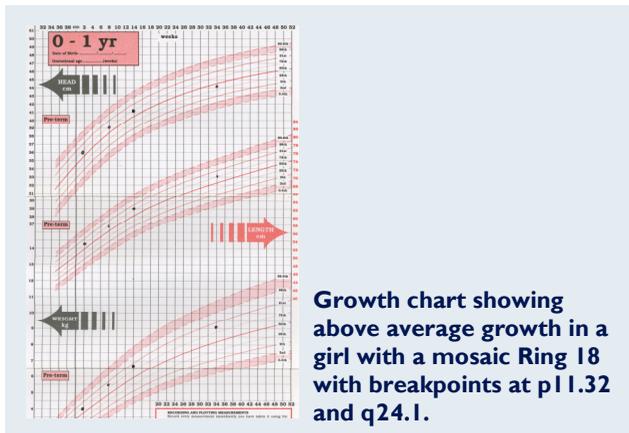
“ Zachary’s head circumference, height and weight were below the 5<sup>th</sup> centile. He has been on growth hormone injections since he was 2 and is now on the 20<sup>th</sup> centile - age 6.

“ Dean’s predicted adult height was up to five foot (150cm). Growth hormone injections appear to have helped his growth spurt to add extra inches to his predicted height - age 17.

Birth statistics recorded at *Unique* suggest that the growth delay does not start in all babies before birth. Among boys, the range of birth weights was between the 9<sup>th</sup> and 70<sup>th</sup> centiles on the standard child growth chart and the range of birth weights for girls was very similar. At around 18 to 20 inches (46 to 51 cm), birth lengths were generally around or slightly below average but within the normal range.

After birth, babies tend to grow slowly and it has been suggested that losing one or more genes from the long arm of chromosome 18 at 18q23 may be one cause of the growth failure although other, as yet unknown, factors also play a role.

Children with Ring 18 are generally tested for growth hormone deficiency and this is sometimes found, with levels varying from normal to severely deficient. Families’ accounts of their child’s treatment with growth hormone are generally positive (Cody 1997; Ghidoni 1997; Hale 2000; U).



## Food and eating

There is little formal information on feeding among babies and children with Ring 18, so much of this information comes from *Unique*. Ring 18 Syndrome can have effects on babies that make feeding more difficult. Babies are often hypotonic and find it hard to suck with enough force to satisfy their own nutritional needs. The floppiness can affect their food pipe (oesophagus) and

contribute to gastro-oesophageal reflux (where feeds return readily from the stomach). Some children also have constipation. Most appear to have a small appetite and almost all need help, either by tube feeding or using a gastrostomy tube to feed direct into the stomach. Babies with a cleft palate need a special bottle or shield until their palate has been repaired. Even babies with a high arch to the palate can find the action of sucking and swallowing difficult. Once babies are weaned, they need to be fed for longer than other children because their relatively immature hand control makes handling finger foods difficult.

These difficulties mean that families should be offered feeding support from the start. If you know that you are expecting a baby with Ring 18 it may be best to make contact with a special needs breastfeeding counsellor during pregnancy to support your attempts to breastfeed. Although some mothers have succeeded in partly breastfeeding their babies, most have not.

## Mobility and hand use

Gross motor development is the development of the large movements a child needs to move independently and reach traditional baby milestones, such as rolling, sitting, shuffling, crawling, walking and running. Fine motor development is using their hands.

Some delay in gross motor development is to be expected but it is difficult to predict the extent. On average *Unique* babies learned to sit between seven and 18 months and children took their first steps between 18 months and five years. Most children need support while walking but eventually most walk independently although this is not possible for all. Some children learn to cycle, play golf, swim, ride ponies, and even try football and badminton. Exercise or swimming classes for children with special needs are helpful and ballet and tap classes can help to improve balance, stability and strength.

Fine motor skills are affected by the floppiness which makes it hard for children with Ring 18 to hold and control a writing implement. Nonetheless, many master a keyboard.

## Families say ...

*“ Feeding was a problem for Tatum because of her cleft palate but now she eats anything and everything - age 3.*

*“ Kiana tried to breastfeed and latched on well but didn't have the muscle to suck, so she failed to thrive and was fed with a squeeze bottle. She had nasal reflux: when she strained to pass a motion, she would reflux out of nose and mouth and would choke - age 5.*

*“ Kate has eaten a varied, healthy diet without many dislikes but has never had a large appetite. She is not greedy, and does not overeat. Reflux was diagnosed at the age of 27 years - age 28.*

## Learning

The formal evidence is that just a few people with Ring 18 will learn at a speed within the normal range. Most will need support for their learning difficulties, but the range is very broad. The majority have a moderate learning disability and a few have a severe disability (Burn 1992; Fryns 1992; Schinzel 2001).

Evidence from *Unique* reflects this broad palette of ability but some individuals have reached an unusually high level of achievement despite their formal learning difficulty. Three teenagers have attained GCSEs (the UK school leaving qualification), in two cases despite moderate learning difficulties. Many families have commented on their child's good memory and their determination to succeed.

Choice of schooling is determined chiefly by local provision. Many children started in a junior school with a unit for the hearing impaired and attended mainstream or special needs secondary schooling.

## Learning - families say ...

- “ Silas is very determined and very inquisitive. He has severe learning difficulties - age 5.*
- “ Zachary can read all the letters of the alphabet, his own and his classmates' names, and write his name. He has an excellent memory and is especially good at academic areas that require memorising. Zachary's learning difficulty is described as mild - age 6.*
- “ Rowy reads magazines and books like Harry Potter and Lord of the Rings. He can write very well but his spelling is basic. He uses Microsoft Office, e-mails and the internet. He has quite a good memory, especially for places and events - age 16, with a moderate learning difficulty.*
- “ Dean reads most things: books, papers, comics, teletext and menus. He writes very slowly and with some degree of difficulty and due to poor muscles in his hands, he tires easily. He can use a keyboard reasonably well and types simple letters and e-mails. He has a very good memory, which is why he reads so well. Reading is probably his most able area - age 18, with a moderate learning difficulty.*
- “ Kate does not read or write but she can manipulate a keyboard with instruction. Despite arthritis, her manual dexterity is good, for example for pricking out plants and sticking small beads. She has a very good memory for roads and places. In a group she has a competitive streak, and is very determined - adult.*

## Speech

Speech is specifically delayed in children with Ring 18 but eventually most children talk, some fluently. There are many reasons for the delay, including the link between the ability to learn and the ability to speak.

Many children with Ring 18 have some hearing loss and those with a cleft or very high palate or enlarged tonsils also have specific difficulties with certain sounds. The early weakness in the muscles of the mouth that showed as insufficient sucking also affects the developing ability to speak.

All children should have speech therapy at first to improve feeding skills and later on to develop the underlying skills for communicating with speech.

Many children are taught to sign and use gestures until their first words emerge, usually in the pre-school and early school years. As with many children with learning difficulties, they need longer to process what they hear and to respond than children without learning difficulties. They frequently have difficulties with specific consonants (see Speech - families say ...).

Children usually make steady progress. Some children, however, communicate fluently, while others do not speak.

## Speech - families say ...

*“Melanie can indicate with gestures and signs and can understand a sentence with several instructions. She has problems with the muscles of her lips, tongue and mouth. Although she can make certain sounds (bababa, ssss, vv), she cannot make p, k or g sounds. She imitates intonations – miaow is a high pitched cry, barking a low pitched cry, small is high pitch and big is low pitch - age 3.*

*“Silas uses signing and gestures to communicate. He has sensorineural hearing loss and is awaiting surgery for cochlear implants. With hand over hand assistance he can sign hi, music, school and done - age 5.*

*“Zachary said his first single words at 4. Now he says phrases like ‘Zac want juice’. He is in the normal intelligence range for non-verbal tasks but his speech is delayed and hard to understand - age 6.*

*“Rowy lipreads and wears hearing aids but has been speaking since he was one. He now uses long and complex sentences - age 16.*

*“Dean started talking at 2 but was unclear, so I taught him Makaton (signing) which helped his speech. He can use long sentences although he tries to get away with the minimum. He understands everything you say but sometimes has a problem knowing how to express himself. He really struggles to communicate with strangers or when he is the focus of attention. He has a higher frequency hearing loss so does not produce the f, s, sh sounds very well - age 17.*

*“Kate communicates by using Makaton signing and gestures and understands a great deal of what is said to her. Her motivation to use signs fluctuates, as does her ability to differentiate and make her own signs specific and recognisable - adult.*

## Medical concerns

### ■ Heart

Losing the end of 18q is known to be associated with heart conditions. Some children are affected while others are not but the association means that all children with Ring 18 will have a thorough cardiac investigation. The information from *Unique* families suggests that heart conditions are often relatively minor, such as a slight murmur, a small hole or an enlargement of the heart muscle and may resolve naturally in time. One child with a breakpoint at 18q22.2 had a large ventricular septal defect (a hole between the lower pumping chambers of the heart) that needed surgery. In most *Unique* members with Ring 18 the breakpoint is close to the tip at 18q23 (U).

### ■ Brain

Loss from either end of chromosome 18 is associated with having a small head and brain (microcephaly); children who have lost material from 18p also have a raised chance of having holoprosencephaly (see page 4). There is some evidence from magnetic resonance images of the brain that myelination (insulation) of the central nervous system is delayed in people with 18q– Syndrome, but the precise meaning of this finding is not certain. Among *Unique*'s 25 members with a Ring 18, one child was reported to have features of holoprosencephaly. Two further children had hydrocephaly, an abnormal increase in the amount of cerebrospinal fluid inside the ventricles of the brain (Gabrielli 1998; Bekiesinska-Figatowska 2001; Linnankivi 2003; U).

### ■ Infections

Many children with Ring 18 have a decreased resistance to infections. This does not affect all children but it means that levels of antibodies in the immune system are routinely checked in some hospitals as typically the levels of an infection-fighting antibody known as IgA (immunoglobulin A) can be low. IgA plays a key role in defending the body against infection that invades through the mucous membranes, such as the nose, eyes, lungs and intestines. It is found in breast milk.

The evidence from *Unique*'s members is that a minority of children do have repeated and quite severe infections.

Parents have reported failure of the stump of the umbilical cord to heal normally, repeated upper and lower respiratory tract infections and viral warts (molluscum contagiosum) (U).

## ■ **Breathing**

The vulnerability to infection means that some babies and children with Ring 18 will have more infections of the lower respiratory tract (chest infections) than other children of their age. The evidence from *Unique's* membership is that although a few people with Ring 18 also develop asthma, this is not typical and is not always linked with the high infection rate.

## ■ **Narrow ear canals**

Loss from 18q is linked with having very narrow or blocked ear canals. Severity varies from narrow ear canals and a small eardrum to absence of the middle ear, blockage of the external ear canal and underdeveloped structures within the inner ear. Both ears are usually affected although one canal may be narrower. A critical region of chromosome loss has been delineated at 18q22.3-18q23.

In *Unique's* experience this is one of the most consistent features of Ring 18, affecting 7 out of 11 children, and this agrees with the findings of researchers of 18q- (64 per cent). Among *Unique's* members, one child has had both ear canals surgically widened and straightened.

Moderate to severe hearing loss is also common and is reported by 9/11 (82 per cent) of *Unique* families. A specific conductive hearing loss caused by temporary blockage in the middle ear (glue ear) or by wax build up in the outer ear is reported by two families but all children with narrow ear canals also have a permanent hearing loss. Most children have been fitted with a bone anchored hearing aid. Hearing aids have been reported to improve hearing although they do not necessarily result in the development of speech (Jayarajan 2000; Nuijten 2003; Veltman 2003; U).

## ■ **Cleft**

People with 18p and 18q deletions have an increased risk of having either a cleft palate (roof of the mouth) or an unusually high arch. Holoprosencephaly (see page 4) often

affects the development of the midline of the face and a cleft can be one result. While a high arch can affect both feeding and speaking (see page 8), a cleft will usually need surgical repair. Among *Unique's* membership, around one third have a high arched palate and half have a cleft. The degree of cleft can range from a small split in the soft palate at the back of the roof of the mouth to a complete gap (U).

### ■ Feet

The feet of babies with Ring 18 are often not perfectly formed. Sometimes feet or toes are very tiny, or one or two toes (usually toes 2 and 3) may cross over each other or be joined by a bridge of skin and tissue. The arch of the foot may be unformed, leaving the sole curved (rocker bottom feet) or the foot may be bent at an unusual angle (talipes, club foot). Many children need surgery and the difficulties often delay the age at which children start to walk (U).

### ■ Eyelids and vision

In a minority of children, loss of material from 18p is linked with ptosis, an inability to fully open the eyelids. Both eyes are usually affected and this is evident at birth and does not develop later. If the ptosis stops your child from seeing properly, it can be corrected in a one or two-stage surgical operation.

Nystagmus (jerky eye movements) is another recognised feature of 18q-. Other than this, although vision appears to be affected in many children and adults with Ring 18, there is no consistent pattern of problems, and it is not clear that they are related to the chromosome disorder. Among vision effects noted by *Unique* members, squint (strabismus) and short sight occur most often. An adult with Ring 18 developed cataracts at the age of 26 (Vogels 2000).

### ■ Genitals

Unusual genital features in boys are known to be linked with loss of material from 18q. In *Unique's* experience this was not common. One boy had hypospadias, where the hole usually sited at the end of the penis is on the underside instead. Depending on how mild this is, it may need no treatment or require corrective surgery to re-site the hole. When this occurs, skin from the foreskin is used so it is normal not to circumcise boys born with hypospadias (U).

## ■ Hands

Thumbs may be positioned unusually low in babies with loss of material from 18q. This feature is sometimes termed 'proximal' thumbs. Some babies also have tapering fingers. A common sign in children with a chromosome disorder including those with Ring 18 is an incurving fifth (little) finger.

## ■ Joints

There is recent evidence that juvenile rheumatoid arthritis is associated with 18q- Eight out of 11 *Unique* families have reported that their children have very loose or hyperextensible joints (Rosen 2004; U).

## ■ Other features

There is a wide range of other features that have been noticed in children and adults with Ring 18. It is uncertain whether they are caused by the loss of material from chromosome 18 or by the general effect of having a ring chromosome or are unconnected with the ring chromosome.

## What therapies is a child likely to need?

Children's needs will be regularly assessed. *Unique's* experience is that children usually need all three key developmental therapies – physiotherapy (physical therapy) to improve their mobility, to develop muscles affected by hypotonia and to help with their large body movements, occupational therapy to help them perform the tasks they will need for everyday living and speech and language therapy to develop their eating, communication and speech. Some families have had access to other therapies such as water therapy (to help mobility), sensory therapy (for behaviour) and early interventions such as portage, a home-visiting educational service for pre-school children. One family has had input from psychology services to help with behaviour.

Information at *Unique* shows that outcomes depend chiefly on children's underlying level of disability, but most children do become mobile and their hypotonia and hand use improve. Level of speech improvement is more variable and is probably closely linked with the child's ability to learn.

## Therapies - families say ...

*“ Occupational therapy from the age of four to help with dressing and so on helped to approach the tasks in a way more constructive to Dean.*

*“ Rowy had speech therapy from age 2 to increase his vocabulary. He had little expressive speech when he began and now speaks clearly and has a wide vocabulary.*

*“ From 5 to 18 Kate had speech therapy to help her communicate. She did not develop speech but her communication skills were helped.*

## Can Ring 18 affect behaviour?

There has been no formal study of the behaviour of people with Ring 18, so we cannot be certain that the behaviour we describe here really reflects something about Ring 18 and does not, for example, reflect an individual's personal characteristics.

Children who cannot express their needs often show frustration by throwing tantrums, screaming, biting or harming themselves and this behaviour eases as they learn to communicate. Meanwhile, management strategies include enhanced versions of everyday parenting techniques: avoiding confrontation, distraction, letting children cry it out, time out and firm holding. As children with chromosome disorders often take longer than expected to process information, some families have commented on the need for patience and consistency in managing difficult behaviour.

A number of families have commented on their child's pleasant and sociable temperament and on their easy pace of life (U).

## Behaviour - families say ...

- “ He saw a psychologist from the age of 7 or 8 to improve his behaviour. This taught me to behave rather than him because I did not really know how to explain things to him or what age to pitch it at.*
- “ Happy and easy going for the most part. Does have temper tantrums, gets frustrated when she doesn't get her own way. Has a hard time understanding NO in reference to dangerous things. Pulls her hair a lot when she gets mad and will also hit her head against a wall if it's available - age 4.*
- “ Very friendly, will go to anybody. Occasionally throws fits, has bitten and hit himself, but this was decreasing by the age of 3. Holding tightly or wrapping up and rocking seems to calm him when he has a fit - age 5.*
- “ Autistic tendencies, likes routines and does not like change. Has anxiety with loud noises. Gets frustrated easily and throws tantrums, screams, throws himself down and kicks and throws objects around - age 6.*
- “ Inappropriate friendliness. Wears her feelings on her sleeve. If she is cross, she lets it out, wherever - age 13.*
- “ Lack of spatial awareness so he sits or stands too close, especially to strangers. Inappropriate friendships – he thinks it's alright to have a 10-year-old girlfriend when he is 15. I have tried to explain this, which is difficult, but the girls of 15 he knows are not interested in being his girlfriend - age 15.*
- “ Happy and easy-going. He is very shy with people he barely knows. He has a very short attention span and needs to be occupied as he tends to meddle with things he shouldn't. He behaves well and is polite. He is just very slow at doing most things and constantly needs pushing to get on with the task. Every day is like Sunday morning to him - age 17.*
- “ Happy, contented, caring, helpful, clever, good at sport - self report from a young adult.*
- “ She has never made close friendships although she always liked to be part of a group. Interested in the rest of her class or group but rebuffs anyone who wants too much attention or becomes too familiar. Great sense of humour (very slapstick) - adult.*

## Will a child ever be able to live independently?

Of *Unique's* two members who have left school, one is living at college and the other is in residential accommodation.

The youngster who is still at college is learning life skills and independence and has his own room where support workers encourage self help and independence skills. He does not yet travel



alone or go out alone to shop but he has a long term girlfriend and dreams of moving to live closer to her home and leaving college to work in a shop.

The adult with Ring 18 shares a sheltered flat with three other wheelchair-dependent adults aged between 20 and 30. After studying life skills and a range of subjects including horticulture, art, drama and information technology at college, she now attends a day centre twice a week, college one day a week and an adult training centre one day. She enjoys TV, going to the pub and bowling and belongs to a church and takes holidays with her carers or her parents.

## Puberty and fertility

The evidence from research is that most people with Ring 18 go through puberty normally at the expected age and that is *Unique's* experience as well, both in young men and women. A number of women with Ring 18 have had children so at least some are fertile (Schinzel 2001; U).

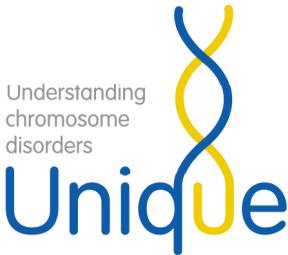
## Can Ring 18 be passed on?

This can happen. There are a number of cases reported in the medical literature, including one mother with learning difficulties who passed a Ring 18 on to her son. There is one report of a completely healthy mother with a mosaic Ring 18 (she had some normal cells and some with Ring 18) who had a son with full Ring 18 (Christensen 1970; Donlan 1986; Fryns 1992).

## Causes

Although a Ring 18 chromosome can occasionally be passed from mother to child, they usually occur out of the blue (*de novo*). Parents are usually offered a test of their own chromosomes but in most cases they are normal. In this case, the chances of having another child affected by a rare chromosome disorder are unlikely to be any higher than for anyone else in the population.

## Support and Information



### **Rare Chromosome Disorder Support Group,**

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[info@rarechromo.org](mailto:info@rarechromo.org) | [www.rarechromo.org](http://www.rarechromo.org)

Join Unique for family links, information and support.

Unique is a charity without government funding, existing entirely on donations and grants. If you can, please make a donation via our website at [www.rarechromo.org](http://www.rarechromo.org) Please help us to help you!

### **The Chromosome 18 Registry & Research Society**

[www.chromosome18.org](http://www.chromosome18.org)

### **The Chromosome 18 Registry & Research Society in Europe**

[www.chromosome18eur.org](http://www.chromosome18eur.org)

Facebook: [www.facebook.com/groups/134997669867413](https://www.facebook.com/groups/134997669867413)

### **Facebook group for Ring 18 in Italian:**

[www.facebook.com/groups/325784750908122](https://www.facebook.com/groups/325784750908122)

Unique lists external message boards and websites in order to be helpful to families looking for information and support. This does not imply that we endorse their content or have any responsibility for it.

This information guide 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. Information on genetic changes is a very fast-moving field and while the information in this guide is believed to be the best available at the time of publication, some facts may later change. Unique does its best to keep abreast of changing information and to review its published guides as needed. The guide was compiled by Unique and reviewed by Dr Jannine Cody, president of the Chromosome 18 Registry November 2004, and by Professor Maj Hultén BSc PhD MD FRCPATH, Professor of Reproductive Genetics, University of Warwick, UK 2005. (PM)

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