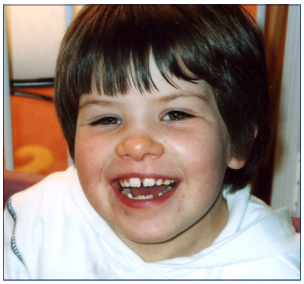
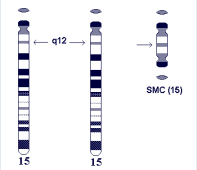
Unique

Idic 15



What is idic 15?

Idic 15 is a chromosome disorder where people have too much genetic material, usually in every cell in their body. In addition to the 46 chromosomes that everyone has, they have a small additional chromosome derived from chromosome 15. Chromosomes come in pairs, and the two normal chromosome 15s are present. The additional chromosome usually consists of two copies of the bit right at the top end of chromosome 15 joined end-to-end in mirror image. The diagram below shows how this works.



Typical features

- Unusual social responses.
- Unusual behaviour.
- 'Floppiness' (low muscletone in early childhood).
- Delay in reaching baby 'milestones'.
- Delay in starting to speak.
- Learning difficulties.

Most children are otherwise healthy. There is marked variation between children and in addition to their unusual chromosome pattern their other genes, personality, environment and opportunities and experiences help to shape their future.

Different names – same condition

Idic (pronounced *eye-dick*) is short for **isodicentric,** meaning that the chromosome contains two copies of the same segment of the chromosome and two of the centromeres ('movement centres').

SMC 15 means **S**upernumerary **M**arker **C**hromosome **15** – an extra chromosome 15. Another name is **Inv Dup 15** (**Inv**erted **Dup**lication **15**)**.**

Some people don't have a separate extra chromosome 15. Instead, one of their chromosome 15s carries a single duplication of the relevant segment, known as **interstitial duplication 15**. This single copy usually causes milder problems than a typical idic 15. Very rarely there is a **triplication**, with two extra copies, and these people, as one might expect, resemble those with idic 15.

Prader-Willi and Angelman (PWACR) critical region

There is a region of chromosome 15 close to the centromere at bands 15q11- q13 with its own name - PWACR, meaning Prader-Willi and Angelman Critical Region. (Prader-Willi and Angelman syndromes are caused by losing one copy of this region.) If the extra copies do not include this region, people rarely face any particular problems and their idic 15 is usually just a harmless family trait. Idic 15 causes more problems if the duplicated fragment includes all or part of this critical region. This leaflet describes the effects on children whose extra copies do include all or part of it.

Will a child look different?

Children with idic 15 do not usually look any different from other children in the school playground. Some parents have remarked that their children are exceptionally good-looking and *Unique* would agree!

Growth is usually normal. A small group of children appear to be unexpectedly short but this is not necessarily caused by their unusual chromosome make-up.

How did this happen?

Chromosome conditions are usually passed down in the sperm and egg cells. This is part of a natural process and as a parent there is nothing you can do to control it. Children from all parts of the world and from all types of background have idic 15. No environmental, dietary or lifestyle factors are known to cause it. So there is nothing you did before you were pregnant or during pregnancy that caused idic 15 to occur and there is nothing you could have done to prevent it.

The extra chromosome material in idic 15 has so far always been found to come from the mother. When egg and sperm cells are formed, the two members of each pair of chromosomes normally line up together and then break and recombine to create new chromosomes that contain different combinations of the genes transmitted by the grandparents to the parents of the child. The recombining can occasionally take place between the wrong broken ends, and you can imagine how this could make an idic 15 chromosome, but this is still a theory as nobody has ever seen it happen.

Sources

This leaflet draws on research referenced in the text and on a survey of *Unique* members and statistical information from *Unique*'s database that included 99 people when the leaflet was written, referenced (U). It also draws on a medical and genetic survey funded by the UK Medical Research Council of 53 people, around half of whom belong to *Unique*. This survey is referenced (1).

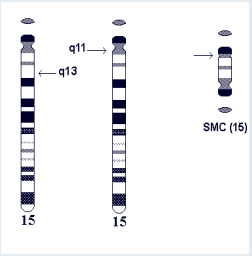
Outlook

Most babies with idic 15 are born perfectly healthy and having idic 15 does not normally make it any more likely that a child will die. The most usual effects of the condition are on behaviour and learning and the only serious medical condition encountered is usually seizures. The oldest person in a recent survey was 57 and *Unique*'s oldest member is in their mid-40s (U).

Supernumerary Marker Chromosome 15

The picture below shows an SMC 15 (a supernumerary marker 15) derived from asymmetrical segments of chromosome 15. One breakpoint is in 15q11, the other is in 15q13.

A fluorescent in situ hybridisation (FISH) study would show that even though the segments are not the same size, each one contains a copy of the Prader-Willi Angelman critical region (PWACR), so this type of SMC15 contains two copies of the PWACR.



How is idic 15 diagnosed?

Idic 15 is diagnosed by examining your child's chromosomes. They are separated from a small blood sample, then examined under a microscope which usually shows the extra chromosome clearly.

An extra genetic test called FISH (fluorescent in situ hybridisation) confirms idic 15 by identifying that the extra chromosome is derived from chromosome 15 and, very importantly, shows whether it contains one or, as is commonly the case, two copies of the PWACR. Extra copies of segments of chromosome 15 that are incorporated into the normal chromosome 15 (duplication or triplication) can be more difficult to see on a routine chromosome study but can be clearly seen with a FISH study or DNA analysis.

Many doctors will not consider a chromosome test important unless a child looks unusual. Babies with idic 15 have no obvious external physical features to suggest they might have a chromosome disorder. Concerns usually only grow after they miss their developmental milestones and even then other more common conditions tend to be considered first. Other explanations offered for the delay in *Unique* children have included being a 'lazy boy' or a 'slow developer'.

This means that families have tended to receive a diagnosis quite late, in many children not until they were three or four years old. The earliest diagnosis known to *Unique* was reached at two months after the parents insisted – contrary to medical opinion - that their baby was strikingly floppy and sleepy.

Does it help to know the exact chromosome pattern?

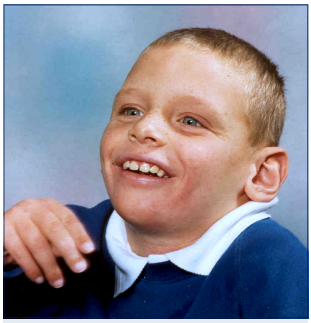
There is a broad range of ability and achievement in children with idic 15. Families who want this are given a description of their child's chromosomes known as a karyotype which will show how much extra chromosome material their child has. But will it help to predict your child's likely future? Doctors are as keen to know as parents and it is an area of active and exciting research.

Three things are known for sure.

- children who have extra material from chromosome 15 that does not contain the critical 15q11- q13 region (PWACR) should normally have no problems at all.
- most children with idic 15 have four copies of the critical 15q11- q13 region. Children with three copies of the critical region show milder effects. Children with five or six copies tend to be more severely affected (Robinson 1993; Browne 1997; Roberts 2002).
- children who have the extra chromosome 15 in only a proportion of their cells also tend to show milder effects, but these could range from complete normality to significant problems. This is a mosaic form of idic 15.

It has recently become clear that people with idic 15 have very variable breakpoints, and that the breakpoint may be different on each of the extra copies of the critical region, so they have two additional copies of most of the genes in the PWACR but one extra copy of some of the genes furthest down the chromosome (Roberts 2003). This variability may help to explain some of the wide variations in children's abilities, but this is not certain.

Research has not yet reached the point where an exact characterisation of your child's chromosomes can act as a roadmap for their future development. Many people doubt that it ever will, because of the huge impact of children's other genes, their personality, environment and the opportunities and experiences offered to them.



How rare is idic 15?

Idic 15 is very rare indeed. No one really knows how many people have it because in a lot of older people it has not been diagnosed. The best estimate suggests that eighteen babies in 100,000 have this extra chromosome.

Idic 15 is actually so uncommon that most doctors and paediatricians have never met an affected child before.

Pregnancy

Pregnancy is usually normal. Two *Unique* babies were born prematurely, but that is as you would expect in any group of babies in the general population.

How can idic 15 affect learning?

Most children need considerable help with learning. In a recent review, researchers found that two out of 13 school age children were at mainstream schools with a statement of special educational need, while the others attended special schools. Among 10 pre-school children, two appeared mildly affected (25).

Among 30 *Unique* members of school age, two children have mild learning difficulties and in five the difficulties are moderate. The other children face a greater degree of learning disability. The children with mild learning difficulties both have a mosaic form of idic 15, and both are fluent speakers. One attended a special needs nursery before transferring to a Steiner-influenced local school with small classes where she is around two years behind her peers. The other girl switched to a special school at the age of 8. Two other children under 8 are in mainstream schools; all other children attend special schools.

Families say …

- *Becky has demonstrated much patience and perseverance at art-based activities. She has demonstrated an excellent memory as she can answer questions on work even when we thought she was not paying attention!* - Becky, age 10.
- *Sarah is like a toddler with 9 years experience – but beside a toddler she is below the most basic skills* - Sarah, age 9.
- *William has a good reading ability and demonstrates good understanding of what he reads. He is trying hard to develop his pencil control. William can match and sequence numbers with ease but numeracy is not an area that interests him* - William, age 8.
- *Alice has had access to an RM Window Box school computer with a touch screen, single switch and Big Mac to exploit simple cause and effect which she does effectively. She is working on tracking programmes and using the touch screen. Alice is sometimes really interested* - Alice, age 7.
- *Tiree is working well to extend her sight vocabulary …. her reading is continuing to develop. She can copy letter shapes .. and when focused can count and order to 10* - Tiree, age 7.
- *Patrick is clever but is just such a busy little boy that he cannot concentrate for very long* - Patrick, age 5.
- *Christian has been estimated by our portage worker to be working at a young year 2 level. Some things he does – puzzles, colour-matching, shape sorters – are only 6 months behind his three years, but his lack of comprehension means that his understanding is only on a simple basis* - Christian, age 3.

Effects on communication

In a recent review of the speech abilities of 33 people aged between 2 and 57, researchers found that all except four children used at least some words. The average age at which children started to speak was 27 months. Two children, both with a mosaic form of idic 15, understood language and spoke normally and seven others could conduct a simple conversation. Seven more used short sentences, five used short phrases and eight children, seven of them under 5 years old, only used single words.

A survey of 30 members of *Unique* aged over 5 found a scattered range of ability. On average *Unique* children first spoke at 3 years 3 months. Seven children speak fluently and while three of these have a mosaic form of idic 15, four have chromosomes with an idic 15 in every cell. Five people, three of them teenagers or pre-teens, speak in sentences, but these are not always clear or easy for people outside the family to understand. Six use single words and 12 children use no words. Both children with five or six rather than four copies of the PWACR region, even in a non-mosaic form, communicate without words. Children communicate by pointing with their eyes, using facial expression, gesture or smiling, laughing or crying. Most lead an adult to what they want and some use a signing or picture exchange system (1; U).

Families say …

- *Claire responded in an appropriate way… her responses were limited, she did not often volunteer information and it was difficult to build a conversation* - from a report on Claire, age 18.
- *Josh uses as little language as possible! He will take our hand and lead us to a toy or employ single words but can string a sentence together if he really wants to* - Josh, age 12.
- *Speech is one of Becky's strong points and she is understood by those who do not know her well. However, the topic of conversation is often of her choosing and unusual, such as 'What do you keep in your shed?'* - Becky, age 10.
- *Robert pulls you or picks up relevant objects like his shoe for going out or a cup for a drink* - Robert, age 9.
- *Olivia has good, albeit delayed expressive language. Her receptive language is much more delayed. This can mislead people who don't know her as she sounds quite 'normal' unless they listen carefully to the content of her speech* - Olivia, age 9.
- *Joshua guides you by the hand. Although he has no speech, Joshua can 'hum' lots of different songs and rhymes and can be very vocal with noises. He can say a few words but they are inconsistent and it can be months before he repeats a word* - Joshua, age 7.
- *Ben uses single words, but understands well in three languages* - Ben, age 5.
- *Sam had an ear for music that was beyond his other developmental levels. He could hum a tune back to you almost perfectly and loved playing instruments* - Sam, age 4.

Effects on mobility and activity

Developmental milestones like rolling over, sitting and walking are delayed but eventually virtually everyone walks independently. *Unique*'s records show a broad range of ages at which children become mobile.

On average children roll at 9 months (range 3 to 24 months). Sitting alone is achieved on average at 14 months (the earliest is 4 months and the latest 3 years and 5 months). Children crawl by 17 months (the earliest is 8 months, the latest 3 years 6 months). First steps come on average at 2 years 2 months (the earliest is 15 months, the latest 4 years 9 months).

Walking alone may seem clumsy at first and your child may have an unusual gait. Some children have difficulties with balance, and others have lax joints.

Hypotonia (low muscle tone) affected 21 out of 37 children in the *Unique* survey (U).

Families say, by the age of 4 ...

- *Over the last year to 18 months Samuel has improved dramatically and now walks, runs, jumps (but not from heights) and climbs (can climb and descend stairs if holding someone's hand and he's focused). He kneels, squats and sits unaided. He loves pushing things along and jumping on his trampoline.*
- *Sven sits in his own way, and crawls like a baby very fast when he wants to get something. He can draw or pull himself to an upright position and tries to walk along furniture. In his mini walker he can 'walk' but it is like pushing forward with his feet. He can walk when I hold his two hands.*

Families say, by the age of 9 ...

- *Will rolls, he cannot sit yet and he is not yet mobile.*
- *Frances runs up to four miles with her Dad and goes on long walks. She learned to cycle at 5, and can ride for miles. She loves jumping into the pool but struggles to swim a width, and finds breathing and leg and arm co-ordination hard. Her ball skills are improving, she can throw a beanbag from one hand to the other. She skips after a fashion.*
- *Although William's gross motor skills appear awkward and he has many difficulties with walking, sitting and standing still, he is able to join in a range of physical movements in PE sessions with some skill! He lacks body awareness and many of his movements can appear impulsive. He has difficulty with balance, both sitting and standing, and finds it very difficult to demonstrate stillness.*
- *Sarah toe-walks and is easily toppled, pedals a trike alone but can't steer and climbs to great heights.*

Medical concerns

Seizures

Seizures are a common feature of idic 15, but they certainly do not affect everyone. In a recent review, fewer than half of 35 people with idic 15 had definitely experienced them (1). In people who had experienced seizures, behaviour and learning abilities tended to be more severely affected. In *Unique*'s own member survey, 22 out of 37 children had experienced seizures. Types of seizure are varied and individual children may experience more than one type. In the *Unique* series, five children have had infantile spasms, a seizure that brings on a cluster of jerks or twitches and is most common in babies between three and 10 months old. Absences – where children stare into space for 5 to 20 seconds – were also common. A scatter of children had other types of seizures including jerks (myoclonic), 'drop attacks' (atonic), stiffening (tonic) or repeated jerking (clonic). 'Grand mal' seizures (tonic-clonic, where the child falls unconscious and then shakes all over) only affected two older teenage members. A small number of children had experienced odd episodes that might have been seizures but had normal EEGs. An EEG (electroencephalogram) is a test that gives a picture of the electrical activity inside the brain.

Some people with idic 15 develop seizures in late childhood or early adolescence. Puberty may also bring a change in the type of seizure. Two families noticed that very hot, changeable or stormy weather was likely to trigger a seizure.
A single antiepileptic medicine or a combination was enough to control the great majority of seizures in *Unique* children and three children outgrew them altogether. In just a few children, seizures proved very hard to control and parents experimented with a variety of alternative treatments. These included yoga, oxygen therapy, craniosacral therapy and homeopathy. The mother of the child treated with homeopathy noted no reduction or intensification of seizures since changing treatment approach but reported that her child's mood, behaviour and concentration improved.

Spinal curvature (scoliosis)

Out of 99 people with idic 15 in *Unique's* records, seven had a noticeable sideways curvature of the spine. This was usually mild but required monitoring. In time and with exercise, the curve usually straightened, but adapted seating was occasionally needed and one *Unique* member had rods inserted into the spine to straighten it.

Frequent infections

It is not clear whether children with idic 15 are more susceptible to infections than other young children. A recent review suggested that they were and that ear infections are especially common. Only seven *Unique* families reported frequent infections and one child was known to have a lowered immune response. Routine immunisations are obviously important (1; U).

Medical concerns - 2

Heart conditions

Six *Unique* children were born with a heart condition, but this usually needed no more than watchful monitoring. One child had an innocent heart murmur, three children were born with a small hole between the two lower chambers of the heart (ventricles, a ventricular septal defect / VSD). This closed naturally in all of them. One child had a persistent ductus arteriosus (PDA, see facing page) which was closed with a clip, and one child had Fallot's tetralogy, a more complex heart condition that does need surgery (see facing page).

Genitals

Out of 57 boys on *Unique*'s database, two had undescended testicles and in two others the hole that is usually at the end of the penis was on the underside instead (hypospadias). This is not a high rate and both of these conditions occur commonly in boys with no chromosome disorder. They can be corrected by a straightforward surgical operation.

Eczema

Dry skin and eczema are extremely common in babies and young children, so it is no surprise to find these conditions in babies with idic 15 and does not imply that the chromosome disorder caused them. Four *Unique* members experienced severe eczema, as did four out of 35 children studied in a recent review. Prescribed emollient and corticosteroid creams kept the condition under control. In one child evening primrose oil was used (I; U).

Puberty

Puberty may not run completely smoothly in girls. One *Unique* member taking hormone injections to halt premature puberty showed cyclical behaviour problems and two girls in a recent review started their periods normally but then experienced scanty or no periods afterwards (1; U).

Hearing

Glue ear causes a fluctuating reduction in hearing and can be treated simply and effectively. *Unique* data suggested that one child in 10 with idic 15 had glue ear; research data suggested that it may be more common (1;U).

Eyesight

Almost one child in five on *Unique*'s records has a confirmed problem with vision, suggesting that all children should have their eyes regularly checked. Apart from strabismus (squint) which is the most common problem, three children have delayed visual maturation, four have some degree of permanent visual impairment and one has a disorder causing impaired central vision (bull's eye maculopathy).

Pain

Children may have a very high pain threshold. This is found frequently in children with chromosome disorders.

Some medical terms

Angelman syndrome A genetic condition where children have seizures, an unsteady walk, a sunny disposition and learning difficulties. It occurs when there is no normally working copy of a gene, *UBE3A,* from the mother. This gene is in the 15q11- q13 critical region and a common cause of the syndrome is loss of the whole region on the chromosome 15 from the mother.

Bull's eye maculopathy A condition that affects the macula, the central part of the retina at the back of the eye that is responsible for highly detailed vision and distinguishing between colours. Children with this condition lose some central vision and have impaired colour vision.

Fallot's tetralogy A combination of two main things. The artery that takes the blood to the lungs has an unusually narrow entrance (pulmonary stenosis). There is also a hole between the two ventricles, the pumping chambers of the heart (ventricular septal defect).

Fundoplication An operation to control gastro oesophageal reflux when medications are not enough. The top of the stomach is wrapped around the bottom of the oesophagus (food pipe) and stitched in place. At the same time, the hole in the diaphragm through which the oesophagus passes is tightened.

Persistent ductus arteriosus The ductus arteriosus (a channel between the aorta and the pulmonary artery that takes blood to the lungs) usually closes shortly after birth. When it stays open, the lungs receive more blood than they should and the heart has to work too hard.

Prader-Willi syndrome A genetic condition where children are short, tend to overeat and become overweight and have learning difficulties. It occurs when there is no copy from the father of genes in the critical 15q11-q13 region, or, occasionally, these genes are present but are switched off.

Enjoyment

Children with idic 15 enjoy just what other children enjoy. Once they are mobile, they love playgrounds and with their lack of fear can climb to heights others would hesitate at. Swimming and horse riding are universal favourites and as their strength, stamina, balance and control improve children extend their activities and some will learn to ride a tricycle or a bicycle, to trampoline, to dance and even to play football. One eleven year old can drop kick - though his grasp of the rules of football is more limited.

Despite their reported autistic tendencies, families consistently say that their children are affectionate and loving.

On average, *Unique* babies smiled at three months.

Effects on behaviour

Babies and young children tend to be quiet and undemanding and do not respond to social cues, although their ability to respond usually increases with maturity.

Among *Unique* families, many parents do note that their babies show a fluctuating interest in other people and are 'in a world' of their own, but they also note laughter and giggles. They live with children who are special because they affectionate, happy and enjoy life. They see their baby maturing out of the passive stage of tiny babyhood into a more assertive toddler stage, with the emergence of temper tantrums and, in some children, challenging and sometimes quite extreme behaviour.

By mid childhood, mood swings and bouts of challenging and angry behaviour may be more evident. They appear to be more common in boys but also occur in girls. Out of the blue, children who have been calm and loving, lash out, pinch, scratch or bite. Three children specifically try to escape. If firm discipline and extremely clear boundaries do not stop the behaviour from escalating, parents must have training in behaviour management or access to psychological support. One family has tried dietary manipulation (gluten-free, low-sugar, added fish oils) with some success. In severe cases, prescribed medicines (such as methylphenidate or risperidone) may be helpful.

By contrast, sensitivity, anxiety and insecurity are big concerns for some families. Children may show extreme anxiety on separation and be easily hurt by other children. Some become extremely anxious when too much is demanded and may hurt themselves (usually biting their wrists or hands) as a result.

Families say …

" *No behaviour problems, a very loving and docile nature. A pussy cat!* - Alice, age 7.

" *Ben is a loving child who wants to be with you and do things together. He joins me in gardening and cleaning and is desperate to talk. The house without him is empty. His impromptu kisses and 'I love you' make up for the moments of hyperactivity! He is very active and demands constant interaction, but can be managed if his attention is engaged appropriately. Generally he is well behaved but he has his moments when he will scratch and bite because he wants to do something he isn't allowed or I don't know what he wants* - Ben, age 5.

" *At regular cyclical intervals for three to five days a month, B can show any, some or all of these behaviours: screaming, shouting, climbing onto furniture, jumping up and down on tables, wetting the floor, flooding the toilet floor and walls, sometimes removing all her clothes, pinching, scratching and headbutting other children, trying to bite herself, removing and throwing glasses, running round the room, trying to get into cupboards. She has no control over this* - Class teacher describing a 9-year-old girl.

Are autistic features common?

A high proportion of children have some autistic traits that may not amount to classical autism but which satisfy formal criteria for an autism spectrum disorder. Many parents have been cautioned by their doctors that their child may develop autistic features. This can be helpful: it took one *Unique* member with an older child three years to achieve the correct diagnosis of autism spectrum disorder for her child, three years that could have been spent instead on appropriate treatment and interventions.

Features may include stereotyped behaviour, no speech or repetitive phrases echoing something in the child's mind or something they have heard, very limited understanding, difficulties with changes in routine and avoiding eye contact. Some families also report obsessive behaviour. However, children with idic 15 show much more sociability than is common in children with autism.

Being alert to early signs means that family support and interventions for the child can start early and some *Unique* members under 5 attend autism pre-schools or follow tailor-made programmes. By mid-childhood most children grow out of behaviour that has been labelled 'autistic' and learn to handle sustained eye contact and social responses more consistently. One child has a formal diagnosis of pathological demand avoidance, a disorder related to autism and Asperger's syndrome. For more information on this, contact the Pathological Demand Avoidance Syndrome Contact Group on www.pdacontact.org.uk.
(Rineer 1998; Wolpert 2000a; Wolpert 2000b; Bolton 2001; U)

Are children with idic 15 hyperactive?

Undoubtedly, some are. Eleven out of 30 families of school age children in the *Unique* survey specifically mentioned hyperactivity. As children become mobile, a high level of restless energy may emerge, especially in boys.

" Ben finds the act of stilling his body to sit for any length of time extremely challenging and needs support to remain seated. I don't think he can actually sit still - Ben, at 5.

Sleep

Sleep problems follow the same patterns as in children without a rare chromosome disorder, but they are more frequent, more extreme and they last for longer. One parent says that the best advice she received was that nearly all sleep problems disappear by the age of 5.
Most families tried the parenting sleep repertoire – routine, relaxation, soothing music, dimmed lights, aromatherapy, massage, gradual withdrawal – with intermittent success. Families who set up clear routines and kept to them found the greatest success, and some families found that sleep problems unexpectedly settled once another issue, often seizure control, control of hyperactivity or reflux medication, was sorted. All the same, for many families respite care proved a godsend and others only took control after sleep training. Four families used prescribed medication. Two families found homeopathy successful.

Independence

A recent review of people with idic 15 included twelve people over the age of 15. In one family there was an unusual idic 15 containing only one copy of the PWACR, identified in a young boy, his mother and his mother's brother. The mother was living independently but needed help from social services with the care of her child; her brother lived in a supervised hostel. Two women without symptoms and living independently whose chromosomes were checked for reasons other than complaints about their own health or development had mosaicism for a typical idic 15 (a mixture of normal and affected cells). The other adults did not live independently and only left home with supervision (1).

Among *Unique*'s members, a 17-year-old was still at school and living at home; a 21-year-old was at a residential college successfully following a course in skills for independent living; a 31-year-old was living at home with his mother.

In terms of personal care skills, many *Unique* members do become completely toilet trained, but some do not achieve this reliably. A few are able to manage without protection during the day by the age of 7, and many are dry and clean in the day time by the time they start secondary school. Almost all are able to co-operate with dressing and feeding and by adolescence many are able to dress with supervision.

What about food and eating?

The *Unique* experience is that while new babies tend to feed slowly and breastfeeding may be hard to establish, early feeding problems are not severe. Some babies have gastro oesophageal reflux – where their feeds flush back up their food pipe and they are either sick or risk aspirating some of the milk into their lungs.

Giving small feeds, positioning your baby semi upright and letting him rest in the same position after a feed may be enough to control reflux but some babies need feed thickeners and antacid medicines to relieve their discomfort. If reflux is very severe, an operation called a fundoplication (see medical terms, p11) can make the valve between the stomach and the foodpipe more efficient.

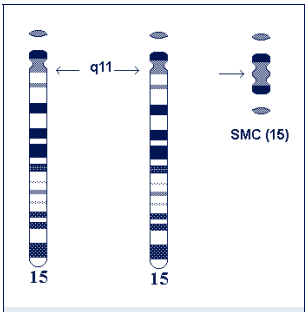
Parents may need to be both patient and ingenious when moving children onto solids. As a rule, children with idic 15 are reluctant to chew and tend to choke on lumps. Your child's speech therapist will be an ally in planning feeding.

Once children start to feed themselves it may become obvious that the sensation of fullness is lacking. Parents report that their child eats regardless of appetite. Some children overfill their mouths until they choke and need their food measured in small quantities. Anticipating this stage should help to avoid some children getting overweight by mid-childhood.

Could this happen again?

The chances of having another child affected by idic 15 depend on the results of chromosome tests on the parents. An idic 15 rearrangement containing the critical 15q11- q13 region usually happens out of the blue and your chances of having another child with idic 15 are no higher than for anyone else in the population.

Very occasionally, a mother who is entirely normal may be mosaic for the idic 15, and then the chance of having another affected child will be significantly raised. Researchers have seen this once in about 50 families investigated (1). The extra chromosome has so far always come from the mother and her age may play a role in some families. Nevertheless, it is probably worth checking both parents' chromosomes.



This SMC 15 has a breakpoint at 15q11. A FISH analysis would show that this type of SMC 15 does not contain any part of the critical region.

The type of idic 15 where the extra chromosome material does **not** contain the critical 15q11-q13 region (*right*) is passed down through families and usually no one is any the wiser until a chance chromosome test shows the abnormality. The only known problem is a possible link with infertility in men.

Each family is unique, so parents of all children with idic 15 should have a chromosome study and a personal interview to discuss the findings with a geneticist or genetic counsellor.

Prenatal diagnosis

Idic 15 can be accurately diagnosed during pregnancy by studying the chromosomes taken either from a chorion villus sample in early pregnancy or from amniotic fluid later on. If you have had a child with idic 15, you may be offered a chromosome test in any future pregnancy. The decision whether to have such a test may be a complex one involving many issues and a discussion with a genetic counsellor may help you to think these through. Don't lose sight of the fact that in nearly all affected families the chance of having another similarly affected child is very low.

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IDEAS

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Unique mentions other organisations' websites to help families looking for information. This does not imply that we endorse their content or have any responsibility for it.

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 Nick Dennis, Wessex Clinical Genetics Service, and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, *Unique*'s chief medical adviser 2004.

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