3q29 deletions and microdeletions
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A 3q29 microdeletion is a rare genetic condition in which a tiny piece is missing from the end of one of the body’s 46 chromosomes. This tiny missing bit increases the possibility of developmental delay, learning difficulties and behaviour problems. However, the effects are often quite subtle and in most people, a small deletion seems to cause no extra medical problems. The subtlety of the effects means that people with the microdeletion may not be diagnosed until quite late.

Chromosomes are the structures in the nucleus of the body’s cells that carry genetic information, telling the body how to develop and function. They come in pairs, one from each parent, and are numbered 1 to 22 approximately from largest to smallest. Each chromosome has a short (p) arm and a long (q) arm.

Looking at 3q29

You can’t see chromosomes with the naked eye, but if you stain them and magnify them under a light microscope, you can see that each one has a distinctive pattern of light and dark bands. You can see these bands in the diagram of the long arm of chromosome 3 on the right.

If you magnify chromosomes about 850 times and look at them down a microscope, a small piece may be visibly missing from near the end of the long arm of chromosome 3. People who have missing material on a chromosome are said to have a deletion but when the amount is so small that it can’t be seen even under a high-powered microscope, it is called a microdeletion.

Larger 3q29 deletions and some microdeletions can be found with conventional chromosome analysis, but other microdeletions can only be found using molecular or DNA technology, in particular a technique using microarrays (array CGH) that shows gains and losses of tiny amounts of DNA throughout the genome and can demonstrate whether particular gene(s) are present or not.

Sources

The information in this guide is drawn from the published medical literature. 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 [www.ncbi.nlm.nih.gov/pubmed]. If you wish, you can obtain articles from Unique.

The guide also draws on information held by Unique. When this guide was updated in 2013, Unique had 32 members with a 3q29 microdeletion, aged between two and 57 years.
**Genetic testing**

Your geneticist or genetic counsellor will be able to tell you about the position and size of the deletion in your child. What your child’s genetic test results show will depend on the technology used. If the chromosomes were examined under a high-powered microscope, the results are usually given as a karyotype, which is likely to read something like this:

46,XY,del(3)(q29q29)dn

- **46** The total number of chromosomes in your child’s cells
- **XY** The two sex chromosomes, XY for males; XX for females
- **del** A deletion, or material is missing
- **3** The deletion is from chromosome 3
- **q29q29** There are two breakpoints in the chromosome, both in band 3q29, indicating a small deletion. If the karyotype says (q29qter) instead, there is one breakpoint, taking out the end of the chromosome
- **dn** The parents’ chromosomes have been checked by analysing a blood sample and no relevant change found at 3q29. The term that geneticists use for this is de novo which means ‘new’.

If array-CGH was used, the results are likely to read something like this:

arr[hg19] 3q29(195,726,835-197,344,663)x1

- **arr** The analysis was by array (arr) comparative genomic hybridisation (cgh)
- **[hg19]** Human genome build 19. This is the reference DNA sequence that the base pair numbers refer to. As more information about the human genome is found, new ‘builds’ of the genome are made and the base pair numbers may be adjusted
- **3q29** The chromosome involved is 3. The position of the deletion is in band 3q29
- **(195,726,835-197,344,663)** The base pairs between 195,719,264 (around 195.7Mb) and 197,344,663 (around 197.3Mb) have been shown to be deleted. Take the first long number from the second and you get 1,625,399 (1.6Mb). This is the number of base pairs that are deleted. **x1** means there is one copy of these base pairs, not two – one on each chromosome 3 – as you would normally expect.

**3q29 microdeletion syndrome**

In the typical 3q29 microdeletion, most people share the same missing segment of around 1.6Mb. The typical microdeletion starts at around 195.7Mb and ends at around 197.3Mb in the 3q29 band. It contains around 20 known genes.

The first published description of a person with a 3q29 microdeletion was in 2001. There have since been around 36 cases reported in the medical literature worldwide. When a particular set of developmental features occurs in a recognisable and consistent pattern in enough people as a result of a single cause, the condition is called a syndrome. The features of a 3q29 microdeletion do occur in this way, so the disorder is often known as 3q29 microdeletion syndrome [Rossi 2001; Willatt 2005; Baynam 2006; Krepischi-Santos 2006; Ballif 2008; Digilio 2009; Li 2009; Tyshchenko 2009; Clayton-Smith 2010; Cobb 2010; Mulle 2010; Quintero-Rivera 2010; Wang 2010; Carroll 2011; Dasouki 2011; Petrin 2011; Sagar 2013].
**Does everyone have the same size deletion?**
No, they don’t. Some people have a larger deletion, others smaller. Yet others have a microdeletion from a different position in the 3q29 band. When the deleted material is from a different position, the effects are likely to be different. Your geneticist or genetic counsellor can tell you the size and position of your child’s deletion.

**How much do we know?**
Comparing different people with the typical 3q29 microdeletion shows that some effects seem to be very broadly similar. This information guide tells you what is known about those effects. Comparing your child’s genetic test results with others, either in the medical literature or within Unique, will 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 test results. It is very important to see your child as an individual and not to make direct comparisons with others with the same results. After all, each of us is unique.

**Are there people with the typical 3q29 microdeletion who have developed normally and have no health or learning difficulties?**
Yes, there are. There are reports in the medical literature of families with a 3q29 microdeletion carried by one of the parents who was themselves healthy and had no suspicion of the deletion until they had an affected child. There are other reports of very mildly affected parents and grandparents (Li 2009; Clayton-Smith 2010; Petrin 2011; Unique).

**Most common features**
Every person with the typical 3q29 microdeletion is unique and so each person will have different medical and developmental concerns. No one person will have all of the features listed in this information guide. However, a number of common features have emerged:
- Usually healthy, without major birth defects
- Some speech delay
- Considerable differences between individuals in terms of learning ability
- In around half, a small head [microcephaly]
- In some cases, a delay in sitting and walking
- Possibly, somewhat similar facial features
- In around one third, features of autism

- **Usually healthy**
Most children and adults seem to be generally healthy. Some children have had frequent ear infections [with the placement of tubes/ grommets in the ear drums to improve hearing] or respiratory infections, including pneumonia, and some have caught more everyday illnesses such as coughs and colds and had difficulty getting over them, but they have outgrown this tendency. Three Unique children with a slightly larger deletion have developed asthma, but this is common in young children [Ballif 2008; Dasouki 2011; Unique]. One 16-year-old boy with a large 3q29 deletion wore hearing aids due to moderate hearing loss [Wang 2010].
Other conditions have only been described in individuals, making it less likely that the 3q29 deletion is the cause of ill health. Three families of children with a slightly larger deletion have reported eczema, but this is very common in young children regardless of whether they have a chromosome disorder. Another child of 10 with a larger deletion developed rheumatic fever; he also developed arthritis at 42 years and has been desensitised for an allergy to bee and wasp stings. A child of four developed a clot in a blood vessel in the brain after a head injury, but this cleared with aspirin treatment [Ballif 2008; Wang 2010; Unique].

■ Usually without major birth defects
The great majority of babies have been born healthy and without major birth defects. Occasionally there is an unusual formation of the chest wall or the spine, most frequently pectus excavatum or pectus carinatum, or a spinal curvature, such as scoliosis. In pectus excavatum, the chest is sunken in (funnel chest). In pectus carinatum, the chest points forward like the breast of a bird (pigeon chest). In some children the chest looks abnormal but this has little effect on heart or lung function. Other children may have breathing difficulties or frequent respiratory infections. The abnormal shape can range from mild to severe and can be progressive. Although this has not been needed for children with a 3q29 deletion, if necessary the chest can be re-shaped in a surgical procedure; the outlook after surgery is generally excellent. If the spine is curved, in many children a slight curve will correct itself in time but progressive scoliosis can lead to problems sitting and if it is severe can cause heart and lung problems. Treatment depends on the severity and progression of the curve but may involve wearing a body brace and surgery to fuse and support the vertebrae [Clayton-Smith 2010; Quintero-Rivera 2010; Dasouki 2011; Unique].

Four/15 boys were born with something unusual about their genitals. Two had hypospadias, where the opening usually at the end of the penis is situated on the underside instead. This is usually repaired in a straightforward operation. One boy was born with undescended testes, corrected surgically at the age of 3, and one had an unusually small, underdeveloped scrotum [Rossi 2001; Ballif 2008; Clayton-Smith 2010]. A heart defect has been found in a few babies. Defects have either been monitored and have resolved naturally in time or with medication, or else were corrected by surgery. Problems vary but the most common is a patent (persistent) ductus arteriosus.
occurs when a short cut between the aorta and the pulmonary artery that takes blood to the lungs fails to close around the time the baby is born. When the ductus arteriosus stays open, the lungs receive more blood than they should and the heart has to work too hard. If it persists in staying open, it can be closed using minimally invasive surgery. Holes between the two upper heart chambers [atrial septal defects/ ASD] were found in at least two babies, one of them closed surgically when the child was 5 years old. Narrowing of one of the key blood vessels leading from the heart was also seen in two babies, in one case affecting the artery that takes blood to the lungs, in the other the aorta that leads from the heart to take blood around the body [Ballif 2008; Digilio 2009; Li 2009; Quintero-Rivera 2010; Dasouki 2011; Unique].

Five babies were born with a cleft [gap] in the roof of their mouth [palate]. Three babies also had a split in their upper lip [cleft lip]. In the other two children, the gap was found in the soft palate at the back of the mouth and in one did not need to be surgically repaired. One of the babies had a particularly large 3q29 deletion, about twice as big as the typical 3q29 microdeletion. A cleft palate causes difficulties both in feeding and in speech production. Surgical repair of the palate eases these difficulties and may eliminate them altogether [Willatt 2005; Wang 2010; Dasouki 2011; Petrin 2011; Unique].

Two children had an inguinal hernia. This shows as a bulge in the area where the lower abdomen meets the upper thigh [the groin]. The cause is that an opening in the lower part of the wall of the abdomen that is open during fetal life but closes before birth does not in fact close. The remaining opening may be small, only allowing fluid through, or it may be large enough for something such as a loop of the intestine or another organ to get stuck in it. An inguinal hernia should always be assessed by your child’s doctors and your child may need surgery to repair it [Li 2009; Dasouki 2011].

Unusual hand and/or foot features. These are usually cosmetic differences, not affecting hand use or walking. Unusual features include long, tapering fingers, curved fingers, and incurving or short fifth fingers. One child had very small feet and in another, the third, fourth and fifth toes curved inwards, while in another the 4th toes curved inwards. Two children had webbing between the toes. Another child had overlapping toes, straightened with physiotherapy. Small, ‘immature’ nails have been observed in two people.

Unusual dental development. Three children had irregularly or widely-spaced teeth and in another, the teeth came through in unusual order. One child had poor tooth enamel [Baynam 2006; Ballif 2008; Quintero-Rivera 2010; Unique].

Some speech delay
The ability to speak and converse appears generally to reflect learning abilities, so children who need greater learning support tend to be those who start speaking later and develop less complex language. Children whose learning ability falls within the normal range may show little or no delay in initially acquiring speech and language, although speech delay may occur in the presence of average cognitive abilities [Cobb 2010]. In children with little or no delay in sitting and walking, a delay in speaking may be the first sign of a developmental problem.

At least one child was found to have dysarthria, a problem articulating the sounds of speech [Quintero-Rivera 2010] and a number of children have been noted to have a nasal or hoarse quality to their voice, in one case attributable to cleft palate surgery [Ballif 2008; Clayton-Smith 2010; Wang 2010].

First words have generally emerged between 17 months and the third year, but delay may
be ongoing and one child was only using two words at eight years. Linked phrases and more fluent speech have emerged between three and seven years, but some children with a severe learning disability have not progressed this far.

The repeated ear infections that some young children with a 3q29 microdeletion suffer bring with them the possibility of a temporary hearing loss caused by fluid within the ear. As children are at risk of speech delay, parental concerns should be acted on early and home or school-based therapy provided. Some children have learned to sign successfully and made the transition from signing to speech.

“ She does not seem to understand what is said and sometimes when asked a question will answer with something unrelated. She uses broken sentences and phrases of 2 to 3 words or more” - 7 years, microdeletion

“At times he uses incorrect words. He finds it hard to join in group conversations and talks excessively, usually on one subject, if engaged in conversation with one person” - 11 years, microdeletion

“Understanding and following conversations is still a problem. She thinks we can read her mind so when she randomly expresses a thought, we should know what it’s in relation to. She has some difficulties with clarity but the family can understand her” - 18 years, deletion

- Considerable differences in terms of learning ability
The range of learning ability is very broad. At one end of the spectrum are adults who gained university qualifications, and children who attend mainstream schools, are able to follow the normal curriculum, sometimes with help for specific learning difficulties, and achieve a range of school-leaving or university qualifications. One child of 6 years was formally tested and found to have average intelligence. At the other end of the spectrum are children and adults with a moderate learning disability. The effect in most children and adults falls between these extremes and is in the mild to moderate range.

Observations at Unique show that specific learning difficulties such as dyslexia can occur and that some children have difficulty concentrating. In terms of schooling, 6/17 children have attended a mainstream [regular] school; the others have attended a special school. In terms of adult outcomes, a number of adults are known to have held down jobs and brought up children, despite in some cases needing extra help at school. This means that any learning difficulties can be extremely mild and the outlook can be good when extra help is given when it is needed [Li 2009; Clayton-Smith 2010; Cobb 2010; Unique].

“She operates at the level of a 2-year-old child and doesn’t have a good memory. But she can teach herself dance moves from music videos and we can see that she learns best when she’s encouraged and praised. She can read single words [such as mum, dad, dog or cat] and has started to write” - 7 years, microdeletion

“He’s level with his age group in English and mathematics and good but not outstanding at art and science. He can read comics and age-appropriate stories and draws with good detail” - 11 years, microdeletion

“She left school at 19 unable to read or tell the time” - adult, deletion

“He has a fairly high verbal IQ but has trouble reasoning and looks ‘slow’. His memory is excellent, he is good at history and keeps up with current events, reading magazines and newspapers” - adult, deletion
In around half, a small head [microcephaly]
50-60 per cent of children with a 3q29 microdeletion have an unusually small head. A baby may be born with a small head or else with an average-sized head, which then grows more slowly than the rest of the body. Occasionally babies are born with an unusually large head. The shape of the head may also be unusual, typically shorter than normal from front to back. In itself, a very small head may have little or no effect, but it is commonly linked with a degree of developmental delay. In one child, a brain scan revealed an abnormal formation of the band of nerve fibres that link the left and right sides of the brain (corpus callosum). In others there was evidence of reduced myelination, the process of creating a layer of insulation around nerve fibres. Myelinated nerves conduct impulses more rapidly than non-myelinated nerves. In other children, brain scans have shown no anomalies [Ballif 2008; Digilio 2009; Clayton-Smith 2010; Quintero-Rivera 2010; Unique].

“Her head is slightly out of proportion to the rest of her body, but not so that people would notice” - 7 years

In some cases, a delay in sitting and walking
Some babies gain head control, sit, crawl and walk at the expected age and grow up into adults with normal mobility and sporting skill. In other babies, some developmental delay occurs. There are marked differences between individuals but it seems likely that a mild delay is most common, with babies sitting independently between seven and nine months and walking by 16 months to three years.

Some babies and children have a degree of low muscle tone and lax joints. In others, some mild spasticity [the joints resist being moved passively] has been seen. Children’s walking style may be unusual [‘ungainly’, ‘stooping’, ‘wide’, ‘plodding’, ‘unsteady’, ‘ataxic’] and individuals have been described with stiff knee and ankle joints that do not fully straighten or have a limited range of movement.

Children with gross motor delay benefit from an early physiotherapy [and possibly occupational therapy] assessment to design a programme of activities and exercises to improve their muscle tone and strength [Ballif 2008; Clayton-Smith 2010; Wang 2010; Unique].

Two children
“She can walk on her own but prefers to hold someone’s hand and seems to stumble and trip a lot. She isn’t swimming yet but she can run and join in play. She loves to dance and joins in aerobics dance DVDs; her coordination seems quite good when doing these” - 7 years, microdeletion

“Normal mobility and gait. Participates in most activities and sports but lacks stamina” - 11 years, microdeletion

Three adults
“No strength to push pedals but can scoot. Loves being on tandem but as we get older it gets less frequent. Very good indeed at bowling” - 18 years, deletion

“She is very stiff and cannot swim or ride a bike but loves to kick a football around. She walks with a wide gait; her legs get very tired after walking” - 20 years, deletion

“He had trouble learning to ride a bike and was not very coordinated, but his coordination has improved and he now rides a bicycle for miles and bowls well” - 50 years, deletion
Possibly, somewhat similar facial features

Children with 3q29 deletion rarely look unusual or ‘different’. There is no typical pattern of features, but among children and adults with the 3q29 microdeletion, features seen most commonly are believed to include a long, narrow face, eyes that slant somewhat downwards, a prominent bridge to the nose, which is typically long with a broad tip, a short philtrum [this is the upper lip between the nose and the mouth] and large ears. Facial asymmetry is also quite common [Clayton-Smith 2010; Quintero-Rivera 2010].

Behaviour: in around one third, features of autism

Unique families’ reports show that children with a 3q29 microdeletion can be loving, happy and communicative. Like other young children, they can also get frustrated, overexcited and irritable. A 3q29 microdeletion appears to increase vulnerability to difficulties with social relationships and communication. Not everyone is affected, but around one in three children have been diagnosed with autism, autistic tendencies or with Asperger syndrome, a form of autism in which there are usually fewer difficulties with learning and language, although social use of language is impaired. Diagnoses have been reached in very young children, although ‘autistic’ behaviours have often been evident long before a diagnosis is reached. Other children are described as having ‘social difficulties’ and in one case social phobia [Wang; 2010]. Behaviours include gaze avoidance, repetitive actions and excessive spinning and jumping, as well as intense and inappropriate anxiety and agitation. Sensory issues are common, including hypersensitivity to sounds, light and touch.

It is not clear if other behaviour difficulties are related specifically to the deletion or generally to the effects of a learning and communication disability. Difficulties with attention/ hyperactivity, poor concentration and limited attention span have all been described. Impulsive behaviour and aggression have been reported, serious enough to lead to school exclusion. Obsessive compulsive behaviour has been described, as have persistent tantrums [Wang 2010].

An association between 3q29 microdeletions and psychiatric disorders seems to be emerging. This does not mean that every child with a 3q29 deletion will develop a psychiatric disorder. It means that as a group, they are more vulnerable to this type of disorder than typically-developing children. In adults or children, depression, bipolar disorder, generalised anxiety, suicidal thoughts, aggression, elective mutism, introversion, panic disorder, schizophrenia and symptoms of psychosis have all been seen, so it is important to watch for early signs. Very unusual behaviour leading to a diagnosis of psychosis has been seen in children aged 5 and 10, so symptoms can present early in life [Willatt 2005; Baynam 2006; Ballif 2008; Digilio 2009; Clayton-Smith 2010; Cobb 2010; Mulle 2010; Quintero-Rivera 2010; Wang 2010; Dasouki 2011; Levinson 2011; Sagar 2013; Unique].

Families need access to consistent support and early intervention if needed. In the UK, these may be provided through a portage scheme or through a Child Development Centre. Later care may be most successful if provided in an environment suitable for those with autism.

“She has difficulties with concentration and tends to copy her sister who is mildly autistic, especially stimming [stereotypy, a repetitive body movement that self stimulates one or more senses in a regulated manner]. She enjoys watching DVDs and dancing and is trying to learn the violin” - child, microdeletion
“He relates normally to family members but his school is organising a social skills course for him” - child, microdeletion

**What else?**

**Development: hand use and coordination (fine motor skills)**

The evidence from Unique is that the development of fine motor skills and hand-eye coordination appears to be relatively spared, with some youngsters able to feed themselves, play, learn personal care skills [dressing, undressing from difficult garments such as bras] and handle implements for drawing and writing on a par with their friends and classmates.

**Growth**

Any effect on growth is unclear. Most babies are born an appropriate weight for the length of pregnancy, although many are born on the low side of normal. One baby in five is small for dates at birth or grows into a child who is short for their age. Any effects on body build are uncertain, but in general it seems that weight is proportionately lower than height, so both children and adults have a tendency to be slight or frankly thin. Among nine adults whose height is known, three men are of normal height, while a fourth is 23cm [9"] shorter than his brothers and 15cm [6"] shorter than his twin sister. Five women are all short, measuring between 147-154cm [4’10”-5’1”] [Digilio 2009; Li 2009; Clayton-Smith 2010; Wang 2010; Dasouki 2011; Unique].

“Short, thin, small head, petite” - child

“She is a good four to five inches shorter than her sisters and has size 3 feet” - adult

**Feeding**

Some babies may have initial feeding difficulties and suck so weakly that they do not meet their own nutritional needs. The evidence from Unique is that these difficulties are not generally severe or long-lasting but mothers and babies may well need intensive early support to establish feeding and breastfeeding. However, out of 9 Unique babies, one needed to be fed through a nasogastric tube, where formula or expressed breast milk is given through a tube threaded through the nose and down to the stomach. Two further babies needed direct feeding through a tube to the stomach [gastrostomy]. Four of the 9 Unique babies and others in the medical literature also had significant reflux, where feeds and stomach contents return into the gullet and are often vomited or may be inhaled, causing chest infections, known as aspiration pneumonia. One of these babies had a surgical operation to treat the reflux and was fed by gastrostomy. Families may also need extra support during weaning [Digilio 2009; Li 2009; Cobb 2010; Dasouki 2011; Unique].

**Outlook**

3q29 microdeletions have only been diagnosed recently. This is not long enough to be certain what the long term effects are. However, as most people have no birth defects and are generally healthy, it seems likely that they will have a normal life expectancy. What is more, some adults were only identified after their children or grandchildren were found to have a 3q29 microdeletion. From the limited information in the medical literature and at Unique, it is clear that some adults are able to hold down a job, have children of their own and make a valuable contribution to their family and, more generally, to society. One point of note is that some adults have experienced significant
mental health problems [Digilio 2009; Li 2009; Clayton-Smith 2010; Unique].

“He lives with his parents and is a great help to them. He is able to cook his meals and take care of the house and dog when his parents are away” - adult

**How did this happen?**
In the majority of people described so far, the 3q29 microdeletion has occurred out of the blue for no obvious reason. The genetic term for this is *de novo* (dn) and a blood test shows that both parents have normal chromosomes. *De novo* 3q29 deletions and microdeletions are caused by a mistake that is thought to occur when the parents’ sperm or egg cells are formed or in the very earliest days after fertilisation.

There are 8 families reported in the medical literature and others at Unique with children who inherited the 3q29 microdeletion from a parent. In one family, the father had the 3q29 microdeletion in a very small proportion of his own cells, but was himself unaffected. Geneticists call this mosaicism, and it can affect any cells in the body [Ballif 2008; Digilio 2009; Li 2009; Clayton-Smith 2010; Petrin 2011; Unique].

What is certain is that as a parent there is nothing you could have done to prevent this from happening. No environmental, dietary, workplace or lifestyle factors are known to cause 3q29 microdeletions. There is nothing that either parent did before or during pregnancy that caused the microdeletion.

**Can this happen again?**
Where both parents have normal chromosomes, it is unlikely that another child will be born with a 3q29 deletion or any other chromosome disorder. Very rarely (less than 1%), both parents have normal chromosomes by a blood test, but a few of their egg or sperm cells carry the 3q29 deletion. This is called *germline mosaicism* and it means that parents whose chromosomes appear normal when their blood is tested can have more than one child with the deletion.

In families where the 3q29 deletion has been inherited from a parent, the possibility of having another child - either a girl or a boy - with the 3q29 deletion rises to 50% in each pregnancy. However, the effect of the microdeletion on the child’s development, health and behaviour cannot be reliably predicted.

Your genetics centre should be able to offer counselling before you have another pregnancy.

**Will my child with a 3q29 deletion have similarly affected children?**
Adults with 3q29 deletions may want to have children. We have not known about the condition for long enough to be certain if it affects fertility but it is likely that fertility will be normal. In each pregnancy, someone with the deletion is likely to have a 50 per cent risk of passing it on and a 50 per cent chance of having a child without the deletion. Their ability to look after a child is very likely to be closely related to any degree of learning difficulty.
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 the original text was reviewed by Dr Lucy Raymond, senior lecturer and consultant in medical genetics, University of Cambridge and Addenbrookes Hospital and by Professor Maj Hultén, Professor of Medical Genetics, University of Warwick, UK. The text has since been updated to include new information.

2013  Version 5 (PM)

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