

UNIQUE
UNDERSTANDING GENES & CHROMOSOMES

2q32 deletions and microdeletions

rarechromo.org

This guide is designed to help families and healthcare professionals looking after people with a 2q32 deletion. It contains information about the cause, the ways in which it can affect people and suggestions about the help and management that can benefit people with this condition.

2q32 deletions and microdeletions

A chromosome 2q32 deletion means that a part of one of the 46 chromosomes has been lost or deleted. If the missing chromosome material contains important instructions for the functioning of the body, developmental delay, some learning difficulties and health problems may occur.

How important these problems are depends on how much of the chromosome has been lost and the location of the deletion.

Genes and chromosomes

Our bodies are made up of billions of cells. Most of the cells contain a set of tens of thousands of genes. Genes act like a set of instructions, controlling our growth and development and how our bodies work. Genes are located on microscopically small, thread-like structures called chromosomes. There are usually 46 chromosomes, 23 inherited from our mother and 23 inherited from our father, so we have two sets of 23 chromosomes in 'pairs'. Apart from two sex chromosomes (two Xs for a girl and an X and a Y for a boy) the chromosomes are numbered 1 to 22, generally from largest to smallest. Chromosome 2 is almost the largest chromosome and contains nearly 1400 known genes. Each chromosome has a short arm (at the top in the diagram on page 3) called p from petit, the French word for small, and a long arm called q (at the bottom). In a 2q deletion, material has been lost from the long arm of one of the two chromosome 2s.

Looking at 2q32

You cannot see chromosomes with the naked eye, but if you stain them and make their image about 1,000 times larger, you can see through a microscope that each one has a pattern of light and dark bands.

In the diagram you can see that the bands are numbered outwards from the point where the long arm meets the short arm (the **centromere**). The 2q32 bands, marked with a red bar, are part way down the long arm and

are divided into 2q32.1, 2q32.2 and 2q32.3.



In a 2q32 deletion, the chromosome has broken in two places usually quite close to each other and the 'sticky' broken ends have rejoined, leaving out the chromosome material between them. This type of deletion is called **interstitial**.

The missing piece of chromosome can be tiny or much larger. If it is large enough to be visible when magnified under a microscope, it is called a **deletion**. Sometimes it is so tiny that it can only be identified using new technology with tests such as FISH or array-CGH. It is then called a **microdeletion**. Smaller deletions generally remove fewer genes. Using the new DNA technologies can show whether particular genes or parts of genes are missing.

The karyotype

Your genetic specialist can tell you more about what chromosome material has been lost. You will often be shown a karyotype, showing the bands where the chromosome has broken and rejoined.

A band can contain many genes and depending on the technology used, the karyotype sometimes shows whether particular genes are present or not. But you will usually need to ask your genetic specialist for a full explanation.

Your child's karyotype may look very like another person's, from Unique or in the medical literature, or it may look exactly the same. But even in people with the same karyotype, the chromosome may have broken at a different point in the same band. This is one important reason why people with apparently similar karyotypes do not all have the same problems or features. Individual differences can be marked and it is important not to make direct comparisons between your child and others. After all, each of us is unique.

But some features and health problems are similar in people with a 2q32 deletion. This leaflet describes the main features of the 2q32 deletion syndrome.

Is there a 2q32 deletion syndrome?

Recently, researchers described four people with similar features and problems and the same deletion from bands 2q32.2 to 2q33. They identified this as the **2q32.2q33 deletion syndrome** (van Buggenhout 2005).

Many other people have parts missing from chromosome 2 that overlap part or all of these bands. These people have some of the same features and problems and some different ones. To identify these people, we searched the medical literature and investigated the Unique database and found reports of 45 people, from newborn babies to adults. One mother passed the deletion on to two of her children (Kreuz 1993).

How many people have a 2q32 microdeletion?

Although larger deletions have been reported for more than 25 years, the technology for detecting 2q32 microdeletions has not been widely available for long, so we don't know yet how common they are. As well as people reported in the medical literature, there are likely to be thousands who remain undiagnosed.

Common Features

Most people with a 2q32.2q33 microdeletion have:

- Developmental delay
- Some learning difficulty or disability
- Slow growth before and after birth
- Severe, lasting feeding difficulties
- High palate or cleft
- Blocked tear ducts
- Hernias in the groin (inguinal)
- Genital anomalies in boys
- Unusual, broad-based walk
- Typical behaviour pattern

Recognisable facial similarities, including: long face, high forehead, small mouth, unusual ears and teeth, thin, sparse and fine hair and thin, transparent skin; a small lower jaw and a small head (microcephaly) (van Buggenhout 2005).

Larger deletions of 2q31q33:

- Small size at birth and growth delay
- Microcephaly
- Cleft palate
- Clouding of the cornea at the front of the eye
- Clenched fingers
- Webbed toes
- Developmental delay and learning difficulties or disabilities

Typical facial appearance including a flat midface, small nose, short, small and widely spaced eyes, sometimes with a hooded upper lid, a thin upper lip, unusual ear shape, small lower jaw, a beaked nose with small external nostrils; also a prominent forehead and a short neck/ redundant neck skin (Mencarelli 2007; Slavotinek 1999; Ramer 1989)

Deletions of 2q24q31:

A picture is emerging from 15 cases: severe epilepsy; low birth weight; postnatal growth delay; learning disabilities; developmental delay; small head (microcephaly); downslanting eyes with long eyelashes; low ears; a small lower jaw (micrognathia); long, broad big toes with a sandal gap and additional foot/ hand anomalies (Pescucci 2007; Boles 1995).

Deletions of 2q33q35:

Low birth weight, growth delay; small head (microcephaly); unusual facial features; developmental delay; cleft palate and small lower jaw (Kramer 2000).

Deletions of 2q33.3q34:

Growth delay before and after birth, developmental delay, learning difficulties/ disabilities, a small head, small lower jaw, unusually formed or low set ears (Courtens 1997).

Will my baby or child look different?

You and the doctors may notice that your baby has an unusual facial appearance. He or she may look more like the other babies and children with a 2q32 deletion than like other members of your family. On page 279 of the van Buggenhout 2005 article (European Journal of Medical Genetics, 48, pp 276-289, available from Unique) you can see four remarkably similar children. Many children, although not all, are fair and have thin, sparse hair, a long face with a high forehead, a small mouth, unusually positioned and formed teeth and unusually formed ears. Their eyes may be unusually short and most typically slant downwards; the upper eyelid may be hooded. The head is typically though not always very small (Mencarelli 2007). Where the deletion covers 2q31 to 2q33, other unusual facial features may be noticed, including a long groove between the nose and upper lip (Ramer 1989). Other features seen occasionally include a prominent forehead, widely spaced eyes, small outer nostrils, and a short neck with sometimes loose skin (Slavotinek 1999).

Typically skin is thin and transparent, with an unuaully thin layer of fat, allowing the blood vessels beneath to show through and there have been reports of people with wrinkly skin over the abdomen and on the backs of the hands and feet as well as an increase in creases on the palms of the hands and the soles of the feet. Hair can be sparse and fine or, by contrast, unusually thick and coarse. These observations, together with the unusually formed teeth (see page 9), suggest that genes in the deleted section of chromosome 2 are important for the usual formation of the skin (ectodermal) and subcutaneous fat layers (Mencarelli 2007; Kreuz 1993).

Hands and feet

In many babies and children, the hands and/or feet have at least some unusual features. The fingers may be clenched, webbed or overlapping, the middle joints may be shortened, some joints may be fused and there may be a single palm crease. When genes from 2q31.1 are within the deleted area, a more serious hand defect may occur with a split between the fingers and growth of only one or two fingers (ectrodactyly) (Bijlsma 2005; Goodman 2002; Slavotinek 1999; Nixon 1997; Boles 1995). Occasionally, the bones of the forearm may be shortened or missing (Bijlsma 2005; Ramer 1990).

Typical features affecting the feet include a sandal gap between the big toe and the second toe; some webbing or joining of some or all of toes 2 to 5 or of some toe joints and a short, broad appearance of these toes; and a long or unusually broad big toe.

It has been suggested that the webbing/ joining of toes 2 to 5 may only occur when genes from 2q31 have been lost and many researchers have suggested that the loss of part or all of a cluster of genes known as HOXD genes at 2q31.1 is the cause of the hand/ foot anomalies. Other 'regulatory' elements outside this 2q31.1 'critical region' must also play a role as people have been identified with hand/ foot anomalies despite having an intact HOXD gene cluster (Pescucci 2007; Svensson 2007; Goodman 2002). One group has suggested that there is a specific gene or genes for the split-hand/foot malformation between the HOXD gene cluster and the centromere (Bijlsma 2005).

In addition to these problems, a significant number of babies are born with unusually angled feet (metatarsus adductus, equinovarus/ clubfoot) for which they will need physiotherapy and probably surgical correction (van Buggenhout 2005; Hiroyama 1990; Ramer 1990; Markovic 1985; Buchanan 1983; Pai 1983; Young 1983; Unique).

Birthweight range at term 2.06kg (4lb 9oz) – 3.7kg (8lb 3oz)

Is there a typical growth pattern?

27/42 showed a slow growth pattern

Babies are often on the small side at birth and continue to grow more slowly than other children, remaining short and thin despite an adequate food intake. Many children have only a thin layer of fat beneath the skin, which contributes to their slim build, and this is sometimes the case even when their height is typical. Height varies, ranging from tall to extremely short – in one case, a nine-year-old was the average height of a four-year-old child. There are few adults diagnosed with a 2q32 deletion but what evidence there is suggests that eventual adult height can be predicted from height as a child, with tall children generally growing into tall adults.

Although a small head (microcephaly) is highly typical of children and adults with a 2q32 deletion, it is not universal and some children appear to follow their familial pattern and have a typical or large head. In one child, the bones of the skull, which are usually separate at birth, fused early, limiting the room for growth of the head and brain. This condition, known as craniosynostosis, will be assessed and if necessary the skull can be remodelled in a surgical procedure (Nixon 1997).

What about food and eating?

Most babies have early difficulties with both sucking and swallowing and need active support with feeding by tube through the nose or in some persistent cases by gastrostomy tube direct into the stomach (Unique). Babies with a cleft palate will experience particular feeding difficulties but the problems are usually obvious even when the palate is intact.

Some babies also experience reflux, in which feeds and stomach contents return up the food passage (oesophagus) and may be vomited or inhaled, with the risk of causing chest infections known as aspiration pneumonia. Careful feeding and positioning both for feeds and sleeping, the use of feed thickeners and medications prescribed to inhibit gastric acid may control reflux. If these measures are not enough, an operation called a fundoplication can be performed to improve the function of the valve from the food passage to the stomach.

Unique publishes a separate guide to Feeding

Development

Most babies and children show a degree of developmental delay and benefit from early intervention with stimulation and play schemes. The extent of any delay varies from child to child and it is better to let your child show his own pace of development than to try to predict it in advance. Your children's centre, developmental paediatrician, opportunity playgroup, portage scheme and health visitor are resources you can turn to for ideas on suitable stimulation.

Toilet training and personal care are typically markedly delayed and many children described in the medical literature have remained fully dependent on others. There is not yet enough evidence from Unique to add to this view.

Unique publishes separate guides to Therapies and Toilet training and continence

How will a child's ability to learn be affected?

Children with a 2q32 deletion can be expected to need some support with their learning but the amount of support needed cannot be predicted just from the karyotype. There are children with a 2q32 deletion who attend mainstream (regular) schools with appropriate support and others, probably the majority, whose needs are better met in an environment that caters for their special needs.

In a number of cases, the ability to learn is affected to a mild or moderate degree and one child with a deletion within the 2q32 bands was reading and understanding language far ahead of his years at the age of 14 years.

This level of achievement will not be possible for all and the researchers who identified the specific 2q32.2 deletion syndrome suggested that most children need considerable support as they have a severe learning disability. Among 12 children whose learning ability was graded, four had a mild level of disability; three had a moderate disability and in five it was severe (van Buggenhout 2005; Gallagher 2003; Unique).

Unique publishes separate guides to Education and Further education, training and work

How will communication be affected?

Children will generally be slower to understand and express themselves in speech and may be helped by learning to sign their needs or by assistive communication systems and devices. In general, it appears that the use of language reflects children's learning ability and those children who face the greatest learning challenges are the least likely to speak or to use more than the occasional word. The typical cleft or high palate also affects speech production, usually making sounds nasal. Researchers have commented on a 'Donald Duck' speech style in two children (van Buggenhout 2005; Unique).

Unique publishes a separate guide to Communication

Sitting, moving: gross motor skills

The major baby milestones of gaining head control, rolling over, sitting, becoming mobile and walking are very likely to be delayed. This means that early physiotherapy (physical therapy) and stimulation programmes should be made available to all children with a 2q32 deletion at the very least as a precautionary measure.

Many babies have a degree of low muscle tone (hypotonia, causing floppiness), but muscle tone may also be raised. Eventual gait may be broad-based and children may walk in short, unsteady steps. Any anomalies of the angle of the foot, such as a 'banana foot' (metatarsus adductus) or club foot (talipes equinovarus) have an obvious impact on mobility but once corrected by surgery or physiotherapy, a smooth walking style should become possible.

There are a number of cases in the medical literature of children with very severely impaired mobility, but this is not directly reflected by Unique, where some babies and toddlers have achieved early milestones within expected limits.

Medical concerns

Cleft palate

Found in 19/44 babies

Typically the palate is high and it may be split (cleft). The hard palate at the front of the mouth may be split or the split may be found further back in the soft, fleshy tissue at the back of the top of the mouth. Occasionally the split is only seen in the tissue that hangs down above the tongue at the very back of the mouth (uvula, known as a bifid uvula when it is split). In some babies with a 2q deletion, a cleft lip may also very occasionally occur. Loss of a gene known as SATB2 from band 2q32 is the most likely cause although children whose deletion does not appear to include the gene have been found with cleft palate (Britanova 2006; van Buggenhout 2005; FitzPatrick 2003; Brewer 1999; Vogels 1997).

Many of the children with a cleft palate have what is known as the Pierre Robin sequence. This combines a cleft palate with an unusually small lower jaw (micrognathia) set back from the upper jaw (retrognathia), frequently leading to feeding and respiratory problems.

The Pierre Robin sequence is most likely to cause difficulties with breathing and feeding in babies. If your baby is affected in this way, the paediatrician and nurses will advise you how to minimise problems. Surgical repair of the palate eases these difficulties and may eliminate them altogether. Occasionally a surgical procedure may be needed to ease breathing and the jaw may be extended using a surgical procedure called a jaw distraction.

As part of the Pierre Robin sequence, the tongue may be displaced backwards towards the throat where it can fall back and obstruct the airway (glossoptosis). Dental development is usually affected by the small jaw and any cleft so that when teeth emerge, they may be small, crooked or missing (see Teeth, page 9).

Heart problems

Found in 8/45 babies or children

Some babies have been born with a heart problem. Large structural defects may be detected on ultrasound scan before birth, but smaller defects may not be identified until the neonatal period or later. Types of problem include holes between the upper or lower chambers of the heart (atrial septal defects (ASD); ventricular septal defects (VSD)). Treatment for an ASD depends on the type, whether the hole closes spontaneously and its size. Treatment can include taking medications to help the heart to work better, control of potential infection to the inner surfaces of the heart and surgical repair with stitches or with a special patch.

Specific treatment for a VSD is determined individually. A baby with a VSD will be evaluated periodically. Small VSDs may close spontaneously; a larger VSD usually needs surgical repair to prevent lung problems that would develop from exposure to extra blood flow.

A number of babies have also been born with a persistent ductus arteriosus. This is a channel between the aorta and the pulmonary artery that takes blood to the lungs which 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. It can be closed using minimally invasive surgery by inserting a coil via an artery in the thigh. Tissue grows around the coil, closing the gap.

Other structural problems have included coarctation of the aorta, in which the blood vessel that takes blood from the heart to the rest of the body is narrowed. This forces the left side of the heart to pump harder. Treatment is tailored to the individual child but if necessary the narrowed section can be surgically removed or made larger (Bijlsma 2005; Maas 2000; Del Campo 1999; Ramer 1990).

Seizures

13/43 people affected

Seizure activity may be apparent early in life and in some babies shows first as apnoea attacks (when babies appear to stop breathing). In other babies the first seizures occur as febrile convulsions with an infection. Seizures may affect the whole brain (generalised) or just part of it (partial) and are typically complex (they affect awareness or consciousness). We don't know yet if there is a typical electrical pattern in the brain of children with a 2q32 deletion who have a seizure. There is quite limited evidence about the course of epilepsy in affected children but it seems that the seizures can be difficult to control and a variety of different medicines are often tried before reasonable control is established (Pescucci 2007; van Buggenhout 2005; Ramer 1990; Unique).

On imaging, various structural anomalies of the brain have been identified, including an absent or underdeveloped corpus callosum (the band of nerve fibres linking the hemispheres), enlarged ventricles (the fluid-filled spaces within the brain) and hypoplasia (underdevelopment) of the cerebellum, the part of the hindbrain that is important for motor and cognitive functions, including time perception, precise movement and learning, particularly unconscious motor tasks like riding a bicycle (van Buggenhout 2005; Ramer 1990; Benson 1986; Buchanan 1983).

Unfortunately, it is not possible to predict which children will develop seizures. Even though imaging of the brain in affected children may show anomalies, since babies and children who experience seizures are more likely to have a brain scan, it isn't possible to make a cause-and-effect link between these findings. Certainly, children with structural anomalies of the brain seem no more likely to develop seizures than those without evidence of a structural change. Similarly, it isn't possible to say that those children with very small heads (microcephaly) are more vulnerable to epilepsy as many babies with microcephaly do not develop seizures and some babies with unusually large heads do (Mencarelli 2007; Unique).

Inguinal hernia

Found in 7/43 babies or children

During fetal development, a boy's testes descend into the scrotum through an opening in the lower part of the wall of the abdomen. The opening usually then closes but if it fails to close or re-opens, fluid or even a loop of the intestine or another organ can pass through. This is an inguinal hernia and it usually appears as a bulge in the groin or as an enlargement of the scrotum. An inguinal hernia should always be assessed by your child's doctors and your child may need surgery to repair it. Although inguinal hernias are more common in boys they can also occur in girls (Mencarelli 2007; van Buggenhout 2005; Loscalzo 2004; Nixon 1997; Ramer 1989; Unique).

Genitalia

Minor unusual features in 11/26 males

A range of minor anomalies of the genitals in boys has been found. This includes small genitals, hypospadias (where the hole usually at the end of the penis is on the underside instead) and undescended testicles, where the testes have not yet completed their natural descent from the abdomen into the scrotum. The approach to undescended testicles depends on the suspected cause but whatever it is, treatment is usually needed if the testicles do not descend naturally in time. If a hormone problem is suspected, a short course of hormone treatment may be suggested. Otherwise, or if hormone treatment does not work, the testicles can be brought down in a short operation under general anaesthetic called an orchidopexy. The scrotum may also be unusually formed or incomplete and a condition known as 'shawl scrotum' has occurred, where the scrotum surrounds the penis like a shawl. In one child with a 2q31 deletion, sex reversal occurred: the external genitals appeared to be female, but the chromosomes were those of a male (Mencarelli 2007; van Buggenhout 2005; Del Campo 1999; Slavotinek 1999; Courtens 1997; Nixon 1997; Young 1983; Unique).

Maturity-onset diabetes

A gene known to be associated with early-onset diabetes (NEUROD1) is found at 2q32, raising the possibility that some people will develop diabetes at a young age. Although this has not yet been shown to occur, parents and health professionals should remain alert to the possibility.

Lung hypoplasia

Very small, underdeveloped lungs were identified in a newborn boy with a 2q33q35 deletion, leaving him dependent on oxygen (Kramer 2000).

Other conditions

A deletion on one chromosome 2 can unmask a condition caused by a faulty gene on the other chromosome 2. This 'double hit' is very unusual but was the cause of a metabolic disorder known as CPSI deficiency in a child with a 2q32q34 deletion (Loscalzo 2004).

Teeth

In some children the teeth grow short, broad and oddly-shaped and there is sometimes a wide gap (known as a diastema) between the top two front teeth. Milk teeth may erupt late and fail to fall out to allow adult teeth through; the set of adult teeth may be incomplete with missing lateral incisors (the teeth on either side of the two front teeth).

It has been suggested that the gene defect underlying children's unusual skin and hair may also affect dental development. Whether that is true or not, children with a 2q32 deletion warrant specialist paediatric dentistry partly because of the genetic influences on their dental development but also because of their early feeding problems (Mencarelli 2007; van Buggenhout 2005; Riegel 2001; Miyazaki 1988; Unique).

Eyesight

The eyes and vision are frequently affected. A developmental defect of the eye known as a coloboma has occurred and underdevelopment of the optic nerve has also been seen, leading to a variable degree of visual impairment.

Corneal clouding and cataract formation has been observed in babies and children with a 2q31q33 deletion and one child had a perforated keratoconus at the age of 16 requiring transplantation of a cornea. Keratoconus is a degenerative condition where the cornea thins and is pushed outwards, usually in the centre, by the internal pressure of the eye. It is a condition which requires to be regularly monitored.

In mild cases, spectacles will offer correction of the refractive problems. Contact lenses may be required for more advanced cases or, as in this case, surgery.

There have also been cases of persistent squint (strabismus) needing surgical correction, and amblyopia, requiring patching to strengthen the weaker, 'lazy' eye (Svensson 2007 ; van Buggenhout 2005; Kramer 2000; Slavotinek 1999; Nixon 1997; Kreuz 1993; Ramer 1990; Narahara 1985; Buchanan 1983; Young 1983; Shabtai 1982; Unique).

In a few cases, the tear ducts have been persistently blocked, requiring opening in a minor surgical operation (van Buggenhout 2005).

General wellbeing

There is no evidence that children with this deletion will be any more unwell than children without a chromosome disorder. Like other young children, they have repeated coughs and colds and are prone to develop ear infections. However, with their precarious weight gain, they may be more likely to stop feeding and lose more weight while ill than other children and to need to spend time in hospital recovering (Unique).

What about behaviour?

A typical pattern of behaviour has been described in some children with a deletion in this region. They have episodes of marked hyperactivity and restlessness and can be chaotic. Although basically they are happy, they can have tantrums inappropriate to their age, outbursts of aggression and anxiety as well as sleep difficulties and some self-harming (headbanging). Not all children are affected and the challenging behaviour can develop at different ages, from 30 months upwards (van Buggenhout 2005; Vogels 1997; Unique).

Researchers have suggested that the underlying cause of the behaviour differences is deletion of a gene known as the GLS gene in 2q32 which plays a role in the production of a neurotransmitter in the brain. This remains to be proven, particularly as children with the typical behaviour pattern have apparently had deletions that did not include this gene. In the meantime, early intervention with behaviour management is strongly recommended for all families before any problems develop. The importance of early intervention is underlined by a description of a youngster with severe problems in the institution where he lives but none at his own home, suggesting that programmes that can support families are important (Mencarelli 2007; van Buggenhout 2005; Vogels 1997).

One boy with a deletion within the 2q32 bands who stopped using previously acquired language at 30 months and developed self injury, tantrums and a dislike of crowds was diagnosed with autism (Gallagher 2003).

> ***He smiles to interact socially. I would say he is borderline shy. He likes to look at new faces, and he smiles at new people, but he does not like for them to carry him. He is also very restless and cannot sit still for more than a couple of minutes" - 2q32.3q33.1 deletion, 22 months***

Unique publishes a separate guide to Challenging Behaviour

Unique publishes a separate guide to Sleep

Why did the 2q32.2 deletion occur?

Some 2q deletions are the result of a rearrangement in one parent's chromosomes. This is usually an inversion in which material has switched directions on one chromosome 2 but no material has been lost or gained and the parent usually has no difficulties with health or development. Occasionally, when the deletion is very small, one parent has the same deletion as the child. A blood test to check the parents' chromosomes will show what the situation is.

Most 2q32 deletions occur when both parents have usual chromosomes. The term that geneticists use for this is **de novo** (**dn**). De novo 2q32 deletions are caused by a change that has usually occurred when the parents' sperm or egg cells were formed. We know that chromosomes must break and rejoin when egg and sperm cells are formed but this only sometimes leads to problems.

Here is one possibility: during the formation of the egg and sperm cells the two members of each pair of chromosomes usually line up together and then break and rejoin to create new chromosomes. These new chromosomes contain different combinations of the genes passed down by the grandparents to the parents of the child. It is believed that after the chromosomes break, the rejoining can take place between the wrong broken ends, and this can lead to a 2q deletion. However, nobody has ever seen this happen, so it is still only an idea.

The breaking and rejoining is part of a natural process and as a parent you cannot change or control it. Children from all parts of the world and from all types of background have 2q deletions. No environmental, dietary or lifestyle factors are known to cause them. There is nothing that either parent did before or during pregnancy that can be shown to have caused the deletion to occur and equally nothing could have been done to prevent it.

Can it happen again?

Where both parents have usual chromosomes, it is unlikely that another child will be born with a 2q32 deletion or any other chromosome disorder. Very rarely (less than 1%), both parents have usual chromosomes by a blood test, but a few of their egg or sperm cells carry the deletion. This is called **germline mosaicism** and it means that parents whose chromosomes appear typical when their blood is tested can have more than one child with the deletion.

Unique publishes a short general guide to Mosaicism

In families where the 2q32 deletion has been inherited from a parent, the possibility of having another child - either a girl or a boy - with the deletion rises to 50% in each pregnancy. However, the effect of the deletion 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 2q32 deletion have similarly affected children?

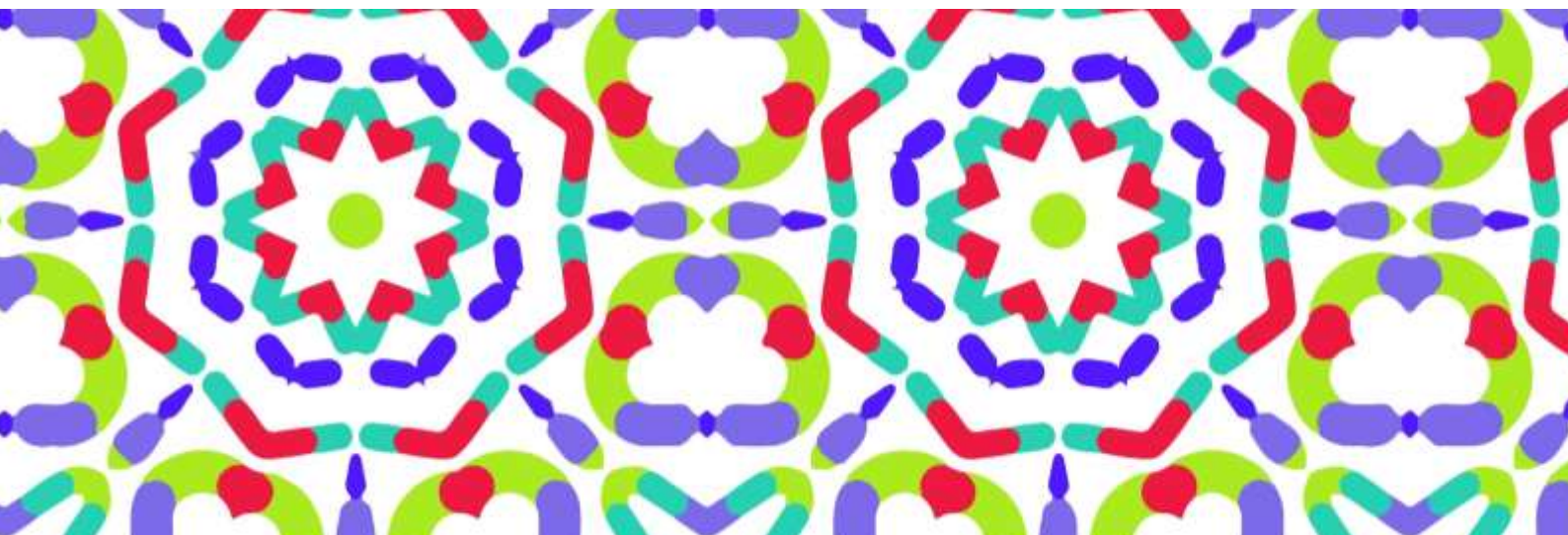
Adults with small 2q32 microdeletions may form close relationships and want to have children. We have not known about the condition for long enough to be certain if it affects fertility but it is certainly possible that fertility will be unaffected. In each pregnancy, someone with the deletion has a 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 their own degree of learning difficulty.

Sources and references

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. If you wish, you can obtain articles from Unique.

The following references contain case reports of individuals with a deletion involving 2q32: Svensson 2007; Mencarelli 2007; Bijlsma 2005; van Buggenhout 2005; Loscalzo 2004; Gallagher 2003; Houdayer 2001; Riegel 2001; Kramer 2000; Del Campo 1999; Slavotinek 1999; Courtens 1997; Vogels 1997; Kreuz 1993; Hiroyama 1990; Palmer 1990; Ramer 1990; Glass 1989; Gorski 1989; Ramer 1989; Dallapiccola 1988; Miyazaki 1988; Sumi 1988; Benson 1986; Markovic 1985; Narahara 1985; Al-Awadi 1983; Buchanan 1983; Franchescini 1983; Pai 1983; Young 1983; Taysi 1981.

The text also includes references to articles discussing deletions near to but not including 2q32. References to Unique are marked Unique.



Inform Network Support

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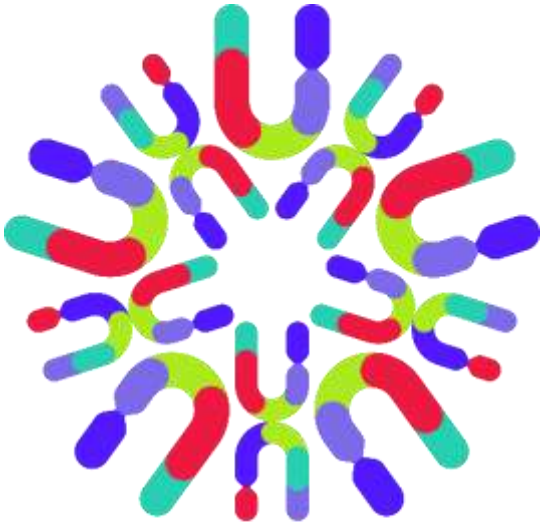
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