

20q13.33 deletions

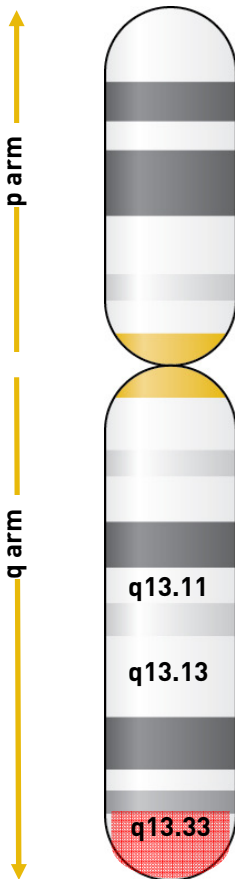


20q13.33 deletions

A 20q13.33 deletion is a rare genetic condition caused by a missing part of one of the body's 46 chromosomes – chromosome 20. For healthy development, chromosomes should contain just the right amount of genetic material – not too much and not too little. A 20q13.33 deletion can result in seizures or epilepsy which can resolve naturally, and developmental delay and/or intellectual disability.

What are chromosomes?

Chromosomes are made up mostly of DNA and are the structures in each of the body's cells that carry the genetic information (in the form of genes) that tells the body how to develop, grow and function. Chromosomes usually come in pairs, with one chromosome from each pair coming from the father and one from the mother.



Of the 46 chromosomes, two are a pair of sex chromosomes, XX (two X chromosomes) in females and XY (one X and one Y chromosome) in males. The remaining 44 chromosomes are grouped in 22 pairs, numbered 1 to 22 from largest to smallest. Chromosomes have a short arm, named p (shown at the top in the figure), and a long arm, named q (shown at the bottom in the figure). The two arms of a chromosome meet at a point called the centromere.

Looking at chromosome 20q13.33

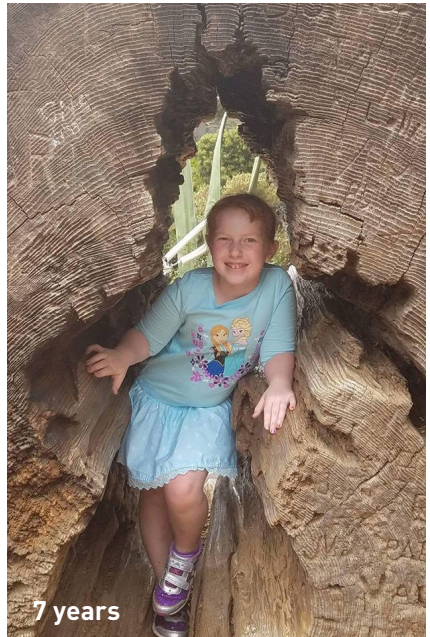
You can't see chromosomes with the naked eye, but if you stain them and magnify them many hundreds of times under a microscope, you can see that each one has a distinctive pattern of light and dark bands.

One of those bands is band 20q13.33. Looking at chromosomes under a microscope, it may be possible to see the genetic material that is missing if the piece is large enough. However, rare chromosome disorders can be caused by subtle changes that are too small to see using a microscope. Molecular DNA technology gives a more precise understanding of the size and position of the deletion. Your geneticist will be able to tell you about the position where the deleted material can be found on the chromosome 20 in your child. More information on molecular DNA technology can be found on pages 12-13.

20q13.33 deletion disorder

A 20q13.33 deletion disorder was first described in the medical literature in 1981. As of 2015, 29 people with a 20q13.33 deletion have been described in the medical literature (see [Sources & references](#) below). Everyone reported in the medical literature had a deletion of all or part of 20q13.33, but the size of the deletion varied. In some children, the deletion included parts of other bands on chromosome 20q: for instance, a deletion starting at band q13.11 and continuing to band q13.33.

Although only 29 people have been reported, that does not mean that there are not more children with a 20q13.33 deletion. There are children registered in international databases, but often with more limited information known. There are also children with a 20q13.33 deletion who have never been included in medical articles.



Main features in children with a 20q13.33 deletion

The features mentioned in this guide have been described in the medical literature in children with a 20q13.33 deletion. It is not known if all features are actually caused by the deletion or whether they occurred coincidentally. Some of the features can also occur in children who don't have the deletion.

Because only a few people with the deletion have been described, not all effects of the deletion are known. The features do vary between children, but one or more of the following features is typical: epilepsy, developmental delay and speech and language delay.

Sources & references

The information in this guide is drawn partly from the published medical literature. With the first-named author and publication date you can look for abstracts or original articles on the internet in PubMed (www.ncbi.nlm.nih.gov/pubmed). When this guide was written, the full text of the articles was used whenever possible, but sometimes only the abstracts were available. As of 2015, 29 people with a 20q13.33 deletion have been described in the medical literature (Fraisie 1981; Aldred 2002; Roberts 2003; Ravnan 2006; Beri-Deixheimer 2007; Bena 2007; Kroepfl 2008; Kurahashi 2009; Traylor 2010; Solomon 2011; Mefford 2012; Pascual 2013; Zara 2013; Allen 2014; Okumura 2015).

When Unique formatted this guide, we had details of 9 members with a 20q13.33 deletion, without any other chromosome change. Four families commented on this guide. Unique contributions are in grey boxes marked with the Unique logo.

How common are 20q13.33 deletions?

It is not known exactly how common the deletion is. As previously stated 29 people have been reported in the medical literature. Furthermore, one person is reported in whom the deletion was present in only a part of the cells of his body. This is called mosaic deletion 20q13.33 (Shabtai 1993).

Outlook

People with the deletion who do not have major health or physical problems are likely to have a normal life expectancy. Three adults with the deletion have been described in the medical literature (Aldred 2002; Zara 2013).

Pregnancy

Most mothers of a child with a 20q13.33 deletion had a normal, uneventful pregnancy. Delivery was normal and they only discovered their baby was affected after the birth.

For 11 children reported in the medical literature information on the pregnancy is included. In three cases no abnormalities were reported (Beri-Deixheimer 2007; Kroepfl 2008). Five children were born via Caesarean section. In one, this was because of pre-eclampsia in the mother (Bena 2007). Another baby was delivered prematurely at 30 weeks after amniotic levels were found to be low, and had respiratory distress (Traylor 2010). Amniotic fluid was low in another pregnancy at 35 weeks gestation, and the baby was in breech position (Traylor 2010). For two pregnancies, no reason is given for the Caesarean section (Traylor 2010; Allen 2014).

Among the other pregnancies, there is one more where amniotic fluid was low (Pascual 2013). One mother went into premature labour (Roberts 2003).

Unique has information on 2 pregnancies. One was uneventful; the other ended in an emergency Caesarean at 33 weeks.



Newborn babies

A newborn with a 20q13.33 deletion does not always show any signs or symptoms of the deletion. But 19 out of 29 newborn babies with the deletion reported in the medical literature had seizures and/or epilepsy which usually started shortly after birth. In one baby, the seizures started as early as four days after delivery. In many babies the seizures resolved spontaneously during early childhood. Medication, when necessary, was usually effective in controlling the epilepsy (Fraisie 1981; Beri-Deixheimer 2007; Kurahashi 2009; Traylor 2010; Mefford 2012; Allen 2013; Pascual 2013; Zara 2013; Okumura 2015). One article describes a 4-year-old girl in whom epilepsy did not respond well to medication (Traylor 2010).

Nineteen out of twenty-nine children may be an overestimation of the frequency of epilepsy in children with a 20q13.33 deletion. This is because in some articles, the presence of epilepsy was the main reason for performing chromosome studies (Kurahashi 2009; Zara 2013).

Researchers think that the epilepsy is caused by the deletion of two genes in particular that are usually deleted in people with a 20q13.33 deletion. These genes are *KCNQ2* and *CHRNA4* (see pages 14-15).

Six/seven Unique children have or have had epilepsy. Seizures started at 5 days in one baby. One girl of 10 years has not had seizures. Brain imaging showed that the band of nerve fibres linking the two sides of the brain (corpus callosum) was missing in one child.



In hospital for EEG testing

“ Our daughter’s epilepsy has been a big battle. She has gone through many medications in her 7 years of life. With some we have had some control, but we eventually lose it or she gets too high on the medication and has to change. This year we had the diagnosis of ESES (Electrical Status Epilepticus of Sleep). This is a severe epilepsy with status seizures occupying nearly all of sleep (must be over 85% for diagnosis) and is associated with cognitive and/or behaviour difficulties. ESES can only be diagnosed with a sleeping EEG (brainwave recording). Families may need to be aware that ESES is possible. Our daughter’s behaviour also seemed to be at its worst when her epilepsy was at its worst.”



Feeding and growth

Feeding difficulties do not appear to be common in babies and children with a 20q13.33 deletion. Two children reported in the medical literature had issues regarding feeding. A boy of 3 years 3 months had failure to thrive, where he could not meet his own nutritional needs. In addition he had hypotonia which means he was floppy. The other boy, nine years old, had difficulties with breast feeding. He was also hypotonic and had motor dyspraxia. Motor dyspraxia is a developmental coordination disorder in which individuals have difficulties performing complex tasks (Traylor 2010). One other child reported showed hypotonia (Mefford 2012).

Some Unique families had significant feeding difficulties. Two comments:

“ Breastfed but with a poor latch leading to poor supply. Seizures had her so exhausted she had no energy to feed properly. She ended up losing weight and wouldn’t take a bottle so had to be fed via nasogastric tube [NG] for a couple of weeks. When weaning she wouldn’t tolerate a spoon in her mouth, or cups, beakers or bottles. She eventually took to solids via baby-led weaning (finger food) and licking food from our finger. She has a good varied diet now but eats very small quantities. She seems to have no natural thirst and has to be prompted to drink fluids.” 4¾ years

“ The diagnosis helped us most with coming to terms with her eating and small size. It was a big struggle and she was unable to take a bottle due to her cleft palate and lack of strength. For 9 months she was fed by NG tube, and only finally came off a prescribed high calorie feed at 9.” 10 years



Growth in children with a 20q13.33 deletion appears to be normal. One exception is a female with skeletal abnormalities reported in the medical literature (see page 9) (Adnan 2002).

Two/3 Unique children are very short. Another is above average height but slim. None is known to have growth hormone deficiency.



Appearance



Most children with a 20q13.33 look like other members of their family. In 10 children reported in the medical literature some distinctive, if variable features in their appearance were noted (Fraise 1981; Aldred 2002; Roberts 2003; Beri-Deixheimer 2007; Kroepfl 2008; Traylor 2010).

Three children had widely spaced eyes (hypertelorism); five children had upslanting eyes, while in one they were downslanting. Two children had an extra fold of skin across the inner corner of their eyes (epicanthal fold). Four children had a bulbous nose and 4 had a thin upper lip. In 3 children the ears were low set or abnormally shaped. Four children had an unusual shape of their skull. In three, the skull showed a temporal indentation, that is, in the part of the skull behind and above the ears. Finally, in one child the distinctive features were unspecified.

Other unusual facial features seen in Unique children include a large or rounded forehead, plagiocephaly (the head is flat at the back or side), and a cleft lip. Two/10 babies, not shown in this guide, were born with a cleft palate, one needing six surgeries to repair it. Two/4 children including one with a cleft palate have a variety of dental problems including late teething, thin enamel and a mouth too small for their teeth.



Development

Fifteen out of 29 children reported in the medical literature had developmental delay and/or intellectual disability (Fraise 1981; Aldred 2002; Roberts 2003; Ravnan 2006; Bena 2007; Beri-Dreixheimer 2007; Kroepfl 2008; Traylor 2010; Mefford 2012; Allen 2014; Okumura 2015). The degree of developmental delay and/or intellectual disability varied from mild to severe.

Sitting, moving and walking

Children may show a delay in reaching their motor developmental milestones. In 7 children described in the medical literature information is given about their motor development. Six of these children sat independently at a mean age of 11.5 months (range 6-21 months). Six children learned to walk at a mean age of

24 months (range 15 months - 3 years, 3 months). A seventh child, 30 months of age, could walk with support at 29 months (Aldred 2002; Roberts 2003; Bena 2007; Beri-Dreixheimer 2007; Kroepfl 2008; Traylor 2010).

One Unique child sat and walked on time. However, she and two other children have difficulties with balance. A girl of 6 cannot jump yet or alternate her feet on stairs.

“ She walked at 22 months with physiotherapy and supports in her shoes/boots. She tires easily. While she is full of energy and often running around non stop, any extra activities take their toll. Walking 2 miles will mean she has no energy later in the day.” 10 years



Learning, communication and speech

Four children with a 20q13.33 deletion are described as having developmental delay or learning difficulties without further details about the severity (Aldred 2002; Ravnan 2006; Traylor 2010; Okumura 2015). Two articles describe children who have mild developmental and language and speech delay. These children had mild attention difficulties (Beri-Dreixheimer 2007; Allen 2013). One boy probably had a severe intellectual disability (Fraisie 1981). Another boy showed normal development at 6 months of age, but needed speech therapy at a later stage (Solomon 2011).

A two-year-old boy had a developmental age of 9-12 months. He was not toilet trained at 7 years of age. One girl was not yet talking at 28 months of age (Beri-Dreixheimer 2007). Another girl was not talking at 30 months of age and had a developmental age of 10 months (Kroepfl 2008). Another boy had a developmental age of 12-15 months when he was 32 months old. He had language delay and spoke some single words. A 4-year-old girl was not toilet trained, could not eat by herself and spoke very few words (Traylor 2010). Yet another girl reportedly had learning difficulties. She spoke in simple sentences at the age of 6 years, 8 months (Bena 2007). A 9-year-old boy with intellectual disability said his first words at 3 years of age and could speak in simple sentences at 5 years of age (Roberts 2003).

An IQ is only reported for one child described in the medical literature: a boy of 9 with an IQ of 40 (Traylor 2010).

Learning difficulties reported to Unique vary in severity but are most typically mild: One child of 4¾ has poor concentration; she had early speech delay which she overcame by 2, but had difficulty following instructions at 3½. A girl of 7 has mild learning difficulties. A girl of 10 is estimated to be 18 months behind her peers in learning. She specifically has great difficulty in remembering what she has heard, and needs multiple repetitions in order to retain information. She did not speak until she was over 3, and used signing until school age. At 10, she has just been discharged from speech therapy.



Behaviour



Behavioural problems have been reported in 6 children with a 20q13.33 deletion. A boy of 3 years, 3 months was hyperactive and sometimes destructive (Traylor 2010). A 4-year-old girl was shy and showed some anxiety (Bena 2007). Three children reportedly made little contact with their surroundings. Two of these children showed autistic features: repetitive behaviour and characteristic hand movements (Beri-Dreixheimer 2007; Kroepfl 2008; Traylor 2010). Another boy was diagnosed with an autistic spectrum disorder (Mefford 2008). A girl with autistic features also had sleeping difficulties (Beri-Dreixheimer 2007).

Three/4 Unique families say their child has a behaviour problem. These include insecurity and anxiety, a sensory processing disorder, finding change hard to handle, and being unable to cope with the demands of everyday life. One family believes that their daughter has PDA (Pathological Demand Avoidance syndrome), a type of autism spectrum disorder.

One child of 10 is pure delight.

“A very happy little girl, She is friendly and caring and is incredibly well liked at her school.”



Medical concerns

■ Head and brain

As mentioned on page 4, epilepsy/seizures appear to occur in a substantial proportion of the children with a 20q13.33 deletion (see [Newborn babies](#), pages 4-5).

Nine children reported in the medical literature had had a CT or MRI of their brain (Beri-Dreixheimer 2007; Kroepfl 2008; Traylor 2010; Mefford 2012; Pascual 2013; Allen 2014). In 4 children results were normal, but in 5 children abnormalities were shown.

One girl had a thin corpus callosum (the brain structure that connects the left and right sides of the brain) (Beri-Dreixheimer 2007). One girl had delayed myelinisation (the natural process of insulating the nerve fibres within the brain). The MRI in another boy also showed this, but results were normal on an MRI performed again at 11 months of age (Mefford 2012). In another girl, the brain ventricles were asymmetrical. In a boy the MRI showed abnormalities in the area surrounding the blood vessels. He had a cystic structure in the left side of his brain (Traylor 2010).

■ Heart

Most children with a 20q13.33 deletion are unlikely to have any heart abnormalities. In three children, heart problems were reported.

A 6-month-old boy had two holes between the lower, pumping chambers of his heart (ventricular septal defect). In a girl of 8 months, the aortic root (part of the main blood vessel leading from the heart to the rest of the body) was underdeveloped. She also had a hole between the upper and lower chambers of her heart. She was born prematurely by Caesarean section after only 30 weeks of pregnancy because there was little amniotic fluid and she showed some respiratory distress. She had several medical problems after birth: a low platelet count, hydronephrosis (low urine flow from the kidney to the bladder) and an infection of her intestines which caused necrosis (necrotising enterocolitis). The necrotising enterocolitis may have partially resulted from her premature birth. Sadly, she died as a result of the infection (Traylor 2010). In a 4-year-old girl, the major blood vessels were wrongly connected to her heart. She had respiratory difficulties as a result (Beri 2007).

All Unique children we know about have a normal heart. One child had a persistent ductus arteriosus (PDA) closed at 16 months. A PDA is a channel between blood vessels leading from the heart that usually closes naturally just after birth.



■ Hands, feet and skeleton

Four children had abnormalities of their hands, feet and/or arms. One child had abnormally curved fingers and skin tissue between the 2nd and 3rd toe. In another child the bones in the fingers and toes were underdeveloped. One boy was born with club feet, a dislocated hip joint, short lower legs and a sacral dimple. He had long, tapering fingers. His lower legs showed movements over which he had no control (Traylor 2010).

Two/10 Unique children have unusual hands or feet. One baby was born with curled in fingers and toes on the right hand and foot; another with toes crossed at birth but evened out by age 10. Her feet are flat and very slim, making it difficult to buy shoes.

“Very sticky soles of feet. Her dad calls them ‘velcro feet’.”



An 18-year-old female had extensive skeletal abnormalities. She was 127 cm (4'2") tall. She had short upper arms and legs and short hands and fingers. She had lordosis (excessive inward curvature of the spine) and both her hip joints were dislocated at birth. These features could point towards a diagnosis of Albright hereditary osteodystrophy (Aldred 2002).

A 68-year-old man who had a mosaic 20q13.33 deletion (the deletion was present in some but not all cells of his body) also had extensive skeletal abnormalities (Shabtai 1993).

One girl had a slightly advanced bone age (Kroepfl 2008).

■ Gastrointestinal problems

Gastrointestinal problems are not reported often in the medical literature in children with a 20q13.33 deletion. One 6-month-old boy had a

tracheoesophageal fistula (a condition in which there is a connection between the windpipe and the oesophagus, the passage that takes food down to the stomach). His oesophagus was underdeveloped (Solomon 2011). Another 9-year-old boy had chronic constipation (Roberts 2003).

The only gastro-intestinal problem we know about at Unique is chronic constipation in a girl of 10, successfully treated with medicines.



■ Eyes and eyesight

One child with a 20q13.33 deletion is blind (Mefford 2012). A four-year-old girl had strabismus (a squint), is longsighted and has an astigmatism (an abnormal curvature of the front of the eye) (Bena 2007). Two children had nystagmus resulting in uncontrolled to and fro eye movements (Kroepfl 2008; Mefford 2012). One boy had ptosis (a drooping upper eyelid) (Traylor 2010).

Two/10 Unique families have reported eyesight problems in their child. One has a squint (strabismus) and ocular albinism, which makes the eyes look paler and affects vision. The other child has a developmental defect of the iris, the coloured part of the eye.



■ Hearing

Hearing difficulties have not been reported in the medical literature in children with a 20q13.33 deletion.

Three/10 Unique children have some degree of hearing loss. One child has hearing loss in one ear; two others have glue ear, a fluctuating deafness very common in young children: one child wears hearing aids for this.



■ Skin

One nine-year-old boy had eczema and a café-au-lait spot (Roberts 2003).

■ Other

Two boys with a 20q13.33 deletion had hypospadias (the hole normally at the end of the penis lies on the underside). One of these boys also had a small scrotum. The other boy had benign cysts in his scrotum (hydrocele and cystic hygroma) (Traylor 2010; Solomon 2011).

One girl had hyperlaxity of her joints (Bena 2007).

Puberty and fertility

There is no information regarding puberty in children with a 20q13.33 deletion as only a few adults with the deletion have been reported. Pubertal development is most likely to be normal; there is one family in which the deletion was passed on through three generations: from a mother to her daughter to her son (Zara 2013). The only feature of the deletion in this family was childhood epilepsy.

Adults with a 20q13.33 deletion

Three adults with a 20q13.33 deletion have been described in the medical literature. Details are only reported for one; a young woman with extensive skeletal abnormalities (see also [Hands, feet and skeleton](#), page 9) and learning difficulties (Aldred 2002).

If one person in a family with the 20q13.33 deletion is mildly affected, will others in the same family also be mildly affected?

It is difficult to answer this question as most people described in the literature are the first and only person in their family with the deletion (de novo deletion, see below). One family is described in which the deletion was present in three generations. The only feature in affected family members was childhood epilepsy (Zara 2013). One father reported passed the deletion on to his daughter, but no information on the father is given (Traylor 2010).

Why did it happen?

When children are conceived the genetic material is copied in the egg and sperm that makes a new child. The biological copying method is not perfect and occasionally random rare changes occur in the genetic code of children that are not seen in the DNA of their parents. The term doctors use for this is [de novo](#). This happens naturally and is not due to your lifestyle or anything you did to cause a change. Most parents of children with a 20q13.33 deletion are not found to carry the deletion. However, one family is described in which the deletion was present in multiple generations. It is therefore important that both parents of a child with the deletion have their own chromosomes tested. In either situation there is nothing you could have done to have stopped this. No one is to blame and nobody is at fault.

Can it happen again?

The chance of having another child affected by a rare chromosome disorder depends on the genetic code of the parent. If the chromosomes in both parents are normal, the chance of having another child affected is very low.

Nonetheless, there is a small chance that part of the egg cells of the mother or part of the sperm cells of the father carry the deletion. You may hear doctors refer to this as [germline mosaicism](#). It means that a blood test that shows the parents do not carry the deletion does not rule out a very small possibility of having another affected child. This has not been reported in the medical literature in 20q13.33 deletions. Another rare possibility is mosaicism. One man who carries the deletion in some but not all of his cells (mosaicism) is reported in the literature. This man had 4 children, but it is not reported if his children inherited the deletion (Shabtai 1993).

The chance of recurrence is much higher if one of the parents is found to carry the deletion or a chromosomal rearrangement that involves chromosome 20. Each family situation is different and a clinical geneticist can give you specific

advice on the chances of recurrence in your family and options for prenatal diagnosis and preimplantation genetic diagnosis (PGD). PGD requires the use of in vitro fertilisation and embryo biopsy, and only healthy embryos are transferred to the mother's uterus. If the parents choose to conceive naturally, prenatal diagnosis options include chorionic villus sampling (CVS) and amniocentesis to test the baby's chromosomes.

Appendix

Results of the chromosome test

In case of a 20q13.33 deletion, the result is likely to read something like one of the following examples:

arr 20q13.33 [59,627,821 – 60,329,092]x1 dn

arr The analysis was performed using the (**arr**) technique: array CGH or SNP array.

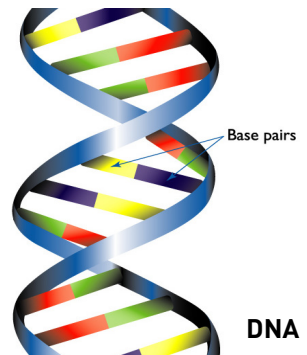
20q13.33 The chromosome involved is chromosome **20** and the band has number **13.33** on the long (**q**) arm.

[59,627,821 – 60,329,092]x1

DNA consists of base pairs. All base pairs are numbers from the top of the chromosome to the end. In this example, the base pairs between **59,627,821** and **60,329,092** are present once (**x1**) instead of the usual twice. This is the deletion. If you subtract the smaller number from the larger number the result is 701,271 (approximately 700,000 base pairs, or 700 Kb, or 0.7Mb).

In summary, doctors call this a 20q13.33 deletion from base pair 59.6 million to 60.3 million (Mb) on chromosome 20 which is 700 kb in size.

dn Short for de novo. See page 12.



1 base pair = bp
1,000 base pairs = 1kb
1,000,000 base pairs = 1Mb

Another example:

ish del (20)(qter)(20qSUBTEL-)

ish This test was performed using a technique called *in situ* hybridisation.
del (20)(qter)

The test showed a **deletion** of chromosome **20**. The deletion involved the last part (terminal part, **qter**) of the long arm (**q**) of chromosome 20.

(20qSUBTEL-)

The technique used a marker for subtelomeres (**SUBTEL**). Telomeres are the last part of a chromosome. A marker is a known piece of DNA. The marker was absent.

Genome Assemblies

The human genome project, an international effort to sequence the entire human genome and map all of its genes, was announced complete in 2003. However, there were many gaps in the sequence and mapping data, and scientists have since been working continuously to identify the missing information. When new sequence information is identified, the base pair numbers of each chromosome change slightly and hence the numbers for individual genes, deletions, and duplications and so on can shift. Sometimes they only shift very little; other times quite a lot.

Each new version of the genome is often referred to as an 'assembly' or a 'build'. Every few years a new assembly is released. The genetic information in this guide is based on the Genome Reference Consortium (GRC) human (h) genome assembly number 37 (GRCh37), which was released in 2009. Confusingly, you will often see the DNA sequence data for this assembly referred to as hg19 (human genome 19) on your/your child's genetic report.

The databases commonly used by clinical geneticists and Unique will soon move to a more recent assembly named GRCh38/hg38, which was released in 2014. Genetic reports will at some point also be altered, so genes and genetic changes may well have new base pair numbers.

Genes



CHRNA4 - Autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE)

KCNQ2 - Benign familial neonatal seizures (BFNS)

Further research involving 20q13.33

The features of a 20q13.33 deletion are likely to be the result of the missing genetic material and missing genes located on this part of the chromosome. It is important, therefore, to have the exact location and length of the missing material determined. When researchers compare the features in different people with the deletion, genes can then be identified that contribute to the different features of 20q13.33 deletions. As a result of earlier research, researchers have established that the KCNQ2 gene and the CHRNA4 gene are probably associated with epilepsy.

KCNQ2 is located within the 20q13.33 band between base pairs 62,037,542 and 62,103,993.

Mutations in the KCNQ2 gene can cause a type of seizure disorder known as benign familial neonatal seizures (BFNS). In this condition, newborns have seizures in the first days after birth. The seizures resolve spontaneously, usually within the first month of life. People with BFNS do have an increased risk (10-15%) of developing epilepsy later in life.

If a gene carries a mutation, the protein that is normally formed can sometimes not function properly. The effect of a mutation is not necessarily the same as the effect of a deletion. Nonetheless, studies among families with BFNS show that, in some, the condition is caused by a deletion of the KCNQ2 gene.

CHRNA4 is located within the 20q13.33 band between base pairs 61,975,420 and 62,009,753.

People with a mutation in the CHRNA4 gene can suffer from a seizure disorder called autosomal dominant nocturnal frontal lobe epilepsy (ADNFLE). In this disorder, people develop seizures during the night. This disorder usually remains present into adulthood.

It is likely that these genes are the cause of epilepsy in children with a 20q13.33 deletion (OMIM; Pascual 2013).

Establishing the role of individual genes is important and can help future research. Often this does not directly result in better treatments. Furthermore, deletion of a gene does not always mean the associated feature is always present. Other genetic and environmental factors also influence this risk.

Support and Information



Understanding
chromosome
disorders

Rare Chromosome Disorder Support Group

The Stables, Station Road West, Oxted, Surrey RH8 9EE
United Kingdom
Tel/Fax: +44(0)1883 723356

info@rarechromo.org | www.rarechromo.org

Unique is a charity without government funding, existing entirely on donations and grants. If you are able to support our work in any way, however small, please make a donation via our website at www.rarechromo.org/html/MakingADonation.asp
Please help us to help you!

20q13.33 Deletion Children is a Facebook group for families:
<https://www.facebook.com/groups/20q13.33/>

This guide was made possible by contributions from: Fonds NutsOhra, Erfocentrum,



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 text was written by Dr Laura van Dussen, MD, Erfocentrum, Netherlands, and Unique and reviewed by Prof Dr C. van Ravenswaaij-Arts (UMC Groningen) and Mieke van Leeuwen (VGnetwerken). With special thanks to Annet van Betuw (VanBetuwAdvies), Marja de Kinderen (PROK Project management and training), Joyce Schaper (Chromosome Foundation) and Sarah Wynn, BSc(Hons) PhD DIC (Unique).

Version 1 hg19 2016. (LvD-PM)

Copyright © Unique 2016