UNIQUE

UNDERSTANDING GENES & CHROMOSOMES

5p deletion syndrome (Cri du Chat syndrome)

rarechromo.org

This guide is designed to help families and healthcare professionals looking after people with 5p deletion syndrome also known as Cri du Chat syndrome. 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.

What is a 5p deletion?

A 5p deletion, also known as Cri du Chat syndrome is a rare genetic condition that can affect health, development and intellectual abilities. The symptoms and features that children and adults with a 5p deletion have are variable and depend on a number of factors including what and how much genetic material is involved as well as each person's own unique genetic background.

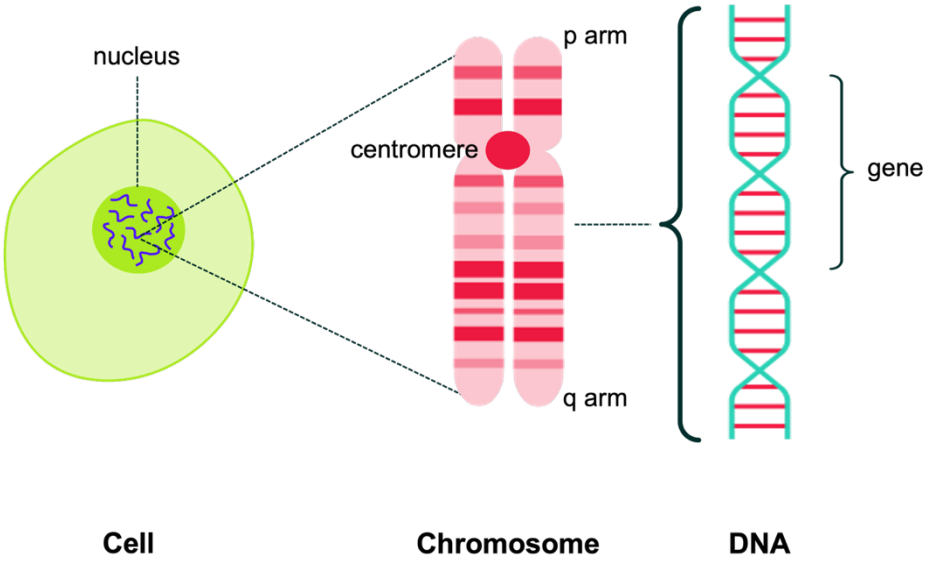
5p deletions were first reported in the 1960s (Lejeune 1963) and some newborns were found to have a distinctive cat-like cry, hence the syndrome's name in French – Cri du Chat, which translates to 'cry of the cat' in English. However, not everyone with this syndrome has this distinctive cry, and this has delayed diagnosis for some people in the past.

What causes a 5p deletion?

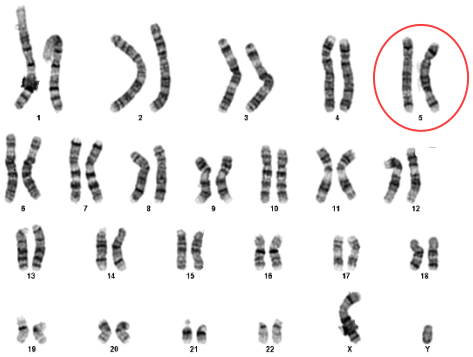
A 5p deletion is caused by the loss of a small piece of genetic material from one of the body's chromosomes – chromosome 5. For typical development, chromosomes should contain the expected amount of genetic material.

Chromosomes, genes and DNA

Our bodies are made up of many different types of cells, almost all of which contain the same set of chromosomes. Each chromosome is made from DNA that codes for our genes. Chromosomes could be thought of as our set of instructional manuals and genes could be thought of as separate sentences or instructions in each manual.



Chromosomes come in pairs with one member of each pair being inherited from each parent. Most of our cells have 23 pairs of chromosomes (a total of 46) as shown in the image below. Chromosome pairs are numbered 1 to 22, roughly according to decreasing size; the 23rd pair comprises the sex chromosomes that determine biological sex. Males usually have one X chromosome and one Y chromosome (XY), and females usually have two X chromosomes (XX).



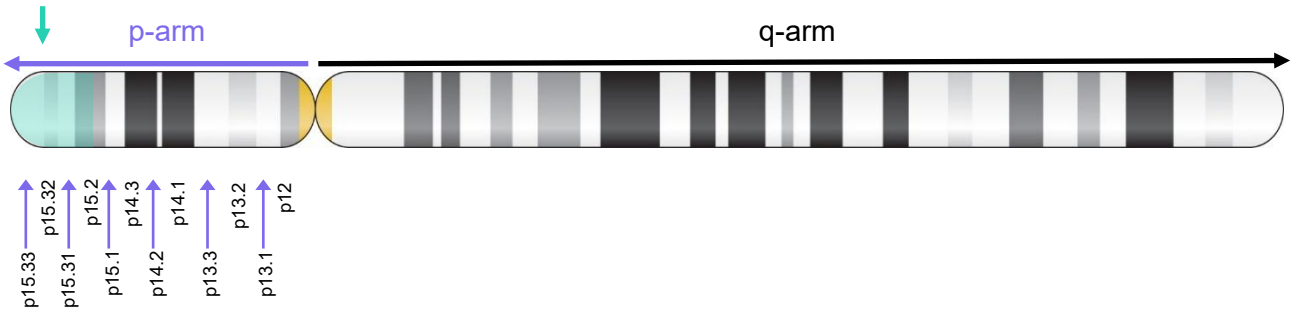
Chromosome pairs 1-22, X and Y (male).
Chromosome pair 5 is circled in red

Chromosomes can't be seen with the naked eye but if cells are prepared in a specific way, the chromosomes can be stained and viewed under a microscope. Each chromosome can be seen to have a specific 'banding pattern' made of light and dark bands. You can see the typical banding pattern for each chromosome in the image opposite, and a more detailed version for chromosome 5 in the image on the following page.

Each chromosome has a short (p) arm and a long (q) arm and the point at which the arms meet is called the centromere (coloured yellow in the image below). Bands are numbered outwards starting from the centromere.

Chromosome 5

Cri du Chat deletion region



5p deletions can occur anywhere in the p-arm of chromosome 5. They vary in size considerably, from very small deletions that only involve one or a few genes, to deletions of the entire p-arm, that involves hundreds of genes. There are also places in the p-arm of chromosome 5 that are prone to breakage, so a number of recurrent deletions of different sizes have been identified; recurrent means that the same or similar deletion has been found in a number of unrelated people.

Each different deletion will contain a different set of genes, the loss of which could contribute to each person's symptoms and features. Deletions associated with Cri du Chat syndrome are not all the same size, but they all involve the end of the p-arm, shaded green in the chromosome image above. These deletions are known as terminal deletions, since they occur at the end (the terminus) of the chromosome. A core set of genes thought to be responsible for the most common symptoms and features associated with Cri du Chat syndrome are thought to be found in this region of 5p. These genes are discussed in further detail on page 15 of this guide.

Cri du Chat syndrome is the most recognized condition associated with a 5p deletion (it is also known as 5p deletion syndrome) but not all individuals with a 5p deletion have Cri du Chat syndrome. People who have deletions that do not involve the bands 5p15.2 to 5p15.3 do not have Cri du Chat syndrome and this guide does not cover such deletions.

Unique publishes a separate guide to deletions and microdeletions

In the past, chromosome deletions were routinely identified by the basic band staining procedure mentioned above. However, many deletions are too small to be seen using this technique. Genetic testing has improved enormously over the years and there are now more precise and detailed tests available such as a chromosome microarray test (CMA, e.g. an array CGH or SNP array) and sequencing (WGS or WES). These types of tests can detect very small deletions even when a specific diagnosis is not suspected. Array and sequencing results provide more precise details of which piece of chromosome is missing and which genes are involved.

Unique publishes separate guides to DNA sequencing, arrayCGH and SNParrays

Genetic Report

Your clinical geneticist or genetic counsellor will have given you detailed information about the piece of chromosome 5 that has been deleted in your child (and perhaps also yourself and/or other family members). The information you are given will include any significant genetic changes that are identified and which significant genes are included in the changes. This will most likely include a chromosome microarray (CMA) test such as that detailed below.

The result of a chromosome microarray test is shown here as an example for a microdeletion within band 5p15.33-p15.1 between base pair (bp) numbers 269,942 and 17,425,722:

arr[hg19] 5p15.33p15.1 (269,942-17,425,722)x1 dn

arr	The analysis was by array (arr)
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. This means base pair positions change depending on the assembly used
5p15.33p15.1	The chromosome involved is chromosome 5 and the position of the deletion is in band p15.33 to p15.1
269,942-17,425,722	The base pairs between 269,942 and 17,425,722 have been shown to be deleted. Take the first long number from the second and you get 17,155,780 (or 17.15 Mb). This is the number of base pairs that are deleted.
x1	Means there is only one copy of these base pairs, not two – one on each chromosome 5 – as you would normally expect, so this a deletion
dn	Means *de novo*. The biological parents' chromosomes have been checked and no deletion or other chromosome change has been found at position 5p15.3p15.1. The deletion is very unlikely to be inherited and has almost certainly occurred for the first time in this family with this child mat here would mean that the deletion has been inherited from the mother; pat here would mean that it has been inherited from the father.

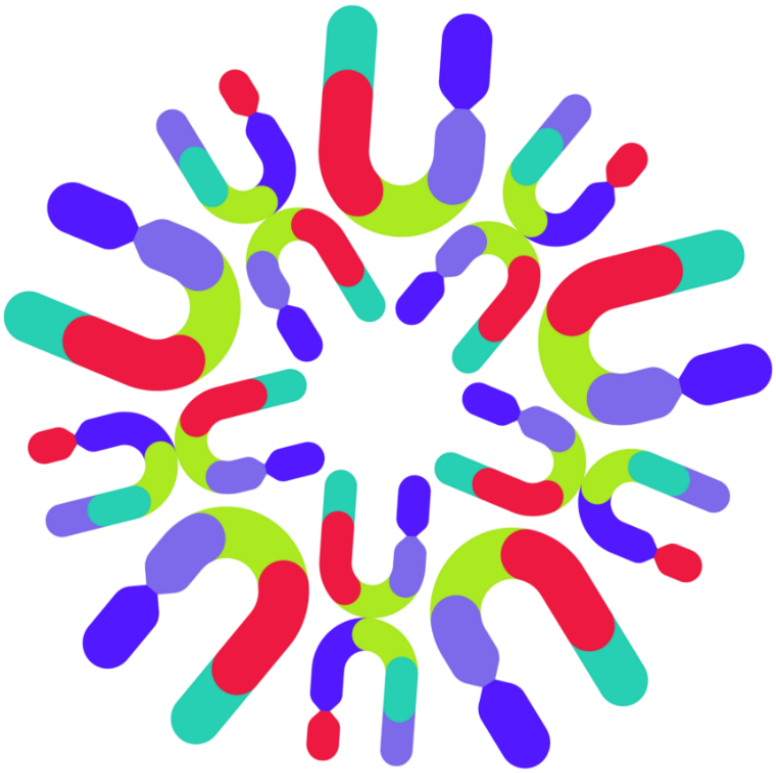
Unique publishes a separate guide to Interpreting Genetic Test Results

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 therefore deletion coordinates can shift.

Each new version of the genome sequence is often referred to as an ‘assembly’. New assemblies are periodically released. The genetic information in this guide is based on the Genome Reference Consortium (GRC) human (h) genome assembly number 38 (GRCh38), the first version of which was released in 2013. The previous assembly was called GRCh37, often also referred to as hg19 (human genome 19) on genetic reports.

It is important to know which reference genome assembly was used to identify the coordinates of a deletion since the coordinates can correspond to slightly different regions of a chromosome depending on which assembly was used.



What features and symptoms do people with a 5p deletion have?

As is common with many genetic conditions, children and adults with a 5p deletion can have a range of symptoms. As more people are diagnosed, and information is shared, the range of symptoms and features, abilities and difficulties and the likelihood of a child or adult having these features will become more clear.

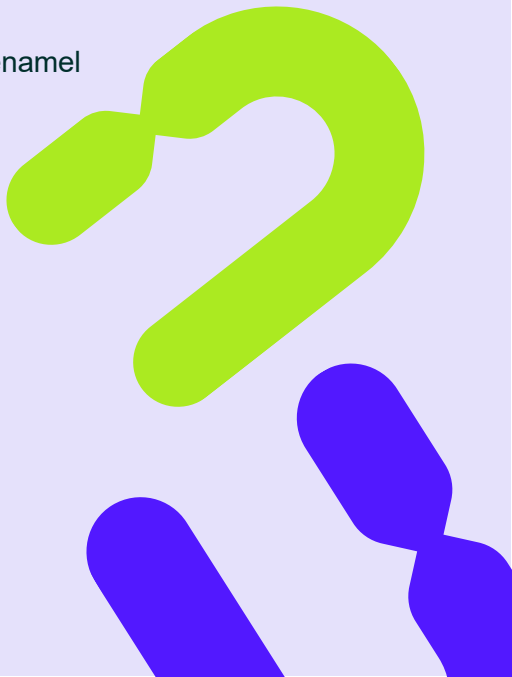
Common features

Children with 5p deletion might have:

- Some degree of developmental delay ranging from mild to profound
- Some degree of intellectual disability (ID) or learning difficulties (LD) ranging from mild to profound
- Speech and language delay or may be non-verbal
- Feeding difficulties which usually resolve after babyhood or early childhood
- Low birth weight
- Gastro-oesophageal reflux (GERD/GORD)
- Constipation
- Characteristic facial features including wide-set eyes, folds on the inner corners of the eyes, flat nasal bridge, low set ears, small jaw, skin tags
- Low muscle tone (hypotonia)
- High muscle tone (hypertonia)
- Joint hypermobility
- Glue ear/frequent ear infections which may resolve during childhood
- Frequent respiratory infections which may resolve during childhood
- Behavioural differences e.g. autistic spectrum disorder (ASD)/ADHD/anxiety
- Small head size (microcephaly)
- Growth delay
- Delayed bone age
- High-arched palate
- Eye/vision anomaly
- Hearing loss
- Spinal curvature e.g. scoliosis
- Skeletal anomaly
- Brain anomaly
- Heart condition which often resolves naturally
- Anomalies of the hands and feet e.g. single crease on the palms, knuckle displacement on the hands, club foot
- Sleep issues
- Dental issues e.g. late teething, over-crowding, tooth-grinding, weak enamel
- Skin condition
- Hernias
- High-pitched cry
- Single crease in palm of hand (single palmer crease)

Other possible features include:

- Early greying of the hair
- A high-pitched, noisy breathing sound (stridor) when breathing
- Seizures
- Short stature but there may be tall stature
- Anomalies of the kidneys and genitals (urogenital anomalies)



Appearance

Certain facial features are found more often in children with a 5p deletion than in other children. These features may mean that you see unexpected similarities between your child and others with a 5p deletion. The most common characteristic features include a round face with wide-set eyes, down-slanting eyes, and skin folds at the inner corner of the eye. Other features include a narrow forehead, a high hairline, low-set or unusually-shaped ears, a nose with a broad bridge, a short space between the nose and upper lip or a small jaw.

Development

Gross and fine motor skills

Developmental delay has been reported in most children with a 5p deletion so far (2025). The degree of delay ranges from mild to profound. Developmental “milestones”, including rolling, sitting, walking, playing with toys, using cutlery, using zips and buttons, and toilet training, are often delayed, although there is a wide range of eventual ability, with some children acquiring mobility and other skills around the same age as “typical” children and others showing more obvious delay. Low muscle tone (hypotonia) is common and may affect mobility. Some children may have an unusual gait when walking because of stiffness, or balance issues (known as ataxia). For some, independent walking may not be achieved. Many benefit from early intervention with treatments or therapies such as orthotics e.g. insoles, braces, splints and callipers; occupational therapy (OT); and physiotherapy (PT).

Unique publishes separate guides to Therapies and Toilet training and continence

Intellectual development and learning

Most children with a 5p deletion have intellectual disability (ID) or learning difficulties. ID ranges from mild to profound but is usually in the severe range and most children have needed additional support with their learning. Early intervention can prove particularly beneficial and formal testing to assess specific, individual needs is recommended.

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

Speech and language

Children with a 5p deletion typically experience some degree of speech and language delay and some may find it difficult to co-ordinate movement of their lips, jaw and tongue to make the right sounds (apraxia of speech). The eventual range of achievement is broad, but many may remain non-verbal. Those who do develop speech may achieve single words, short phrases or basic sentences and a few go on to develop conversational skills and a broad vocabulary. Many parents believe that their child can understand a lot more than they can express.

An assessment by a speech therapist should be able to identify your child’s specific difficulties, allowing regular therapy sessions tailored to your child’s specific areas of need. Where individuals have no speech or very few words, Augmentative and Alternative

Communication (AAC) methods, including pointing, pictograms, gestures, facial expression and simplified sign language and high-tech communication systems (aided communication) have enabled many to communicate their thoughts and needs well.

“At 5 our son still had the cat cry and had no meaningful conversation but now at 41, although he has no verbal language, he has a massive vocabulary and uses key word signing and his electronic voice aid.”

Unique publishes a separate guide to Communication

Feeding

Feeding issues in the new-born period are common. Low muscle tone may contribute to difficulties with swallowing and some babies will suck weakly and may need high energy milks to encourage weight gain. Many babies also suffer from gastro-oesophageal reflux (GERD/GORD) (in which feeds return readily up the food passage), which may require treatment, including careful positioning for feeds, medication, nutritional supplements or, in some cases, insertion of a nasogastric tube (NGT) or percutaneous endoscopic gastrostomy tube (PEG/G-tube). Other issues that have been reported include aspiration (where fluid, food or saliva enters the airway or lungs) or a strong dislike of having anything in or around the mouth (oral aversion). Some children have benefited from attending a feeding clinic where an assessment can be made, and advice to help treat any eating and drinking difficulties provided.

Unique publishes a separate guide to Feeding

Constipation

Constipation is common among children with a 5p deletion and can be related to low muscle tone, little exercise, a low-bulk diet and small fluid intake, among other reasons that are not fully understood. It is important that you discuss the possible causes with their health visitor or doctor, who may recommend adapting your child’s diet or giving stool softeners or laxatives. Some children have benefitted from enemas when symptoms were particularly severe.

Growth and stature

Most children with a 5p deletion described in the medical literature so far (2025) are noted as having growth delay and short stature. Most of these children also have smaller than expected head size (microcephaly). Their bone age is also delayed compared to their actual (chronological) age. Beyond infancy, height and weight often remains below average. However, some individuals have tall stature. There are specific growth charts available for children who have a 5p deletion (Marinescu 2000). What may be considered small for expected age might be average for an individual with a 5p deletion.

Behaviour

Children with a 5p deletion typically tend to have behaviour in keeping with their overall degree of developmental delay, and most have a happy disposition. Some children have an autism spectrum disorder (ASD) diagnosis or autistic traits. Other behaviours including attention deficit hyperactivity disorder (ADHD), anxiety, self-harming, aggressive behaviours, obsessive compulsive disorder (OCD), or impulsivity have also been reported.

Some children also have sleep problems. Children usually benefit from consistent routines, boundaries, rewards and other behaviour management techniques. Efforts to take into account and introduce strategies to tackle communication and other difficulties can also be beneficial.

Unique publishes a separate guide to Challenging Behaviour

Sleep

Sleep problems are a common concern for families. These can include difficulty falling asleep, frequent night wakings, and unusual breathing patterns during sleep, such as sleep apnoea (where breathing temporarily stops). If sleep apnoea is suspected due to snoring or observed pauses in breathing, an evaluation by a specialist and a sleep study may be recommended.

Unique publishes a separate guide to Sleep

Puberty

There is limited information available about puberty in children with a 5p deletion. We do know that most boys and girls appeared to go through puberty as expected or mildly delayed. Some families of children with chromosome disorders and behavioural or learning difficulties can be particularly concerned at their daughter's ability to cope with menstruation, and for some discussing menstrual regulation options with a paediatrician may be beneficial.

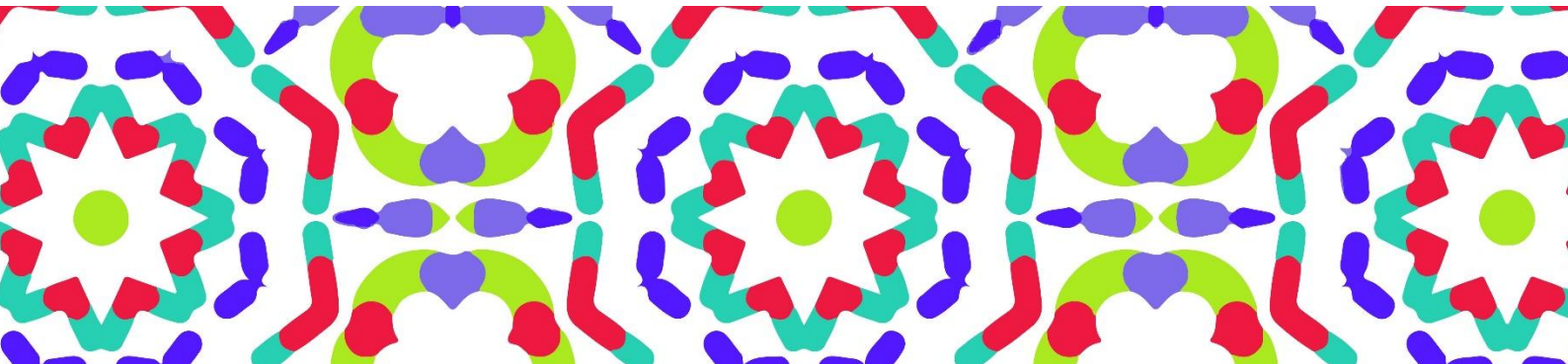
Unique publishes a separate guide to Puberty

Adulthood

Information on adulthood for people with a 5p deletion is still emerging. Experiences are likely to vary considerably depending on an individual's intellectual abilities and any ongoing medical concerns. Adults will have varying levels of independence, while some will live independently, others with greater intellectual difficulties may continue to live with their parents or in supported settings such as a group or residential care home. For adults whose ID is severe, it is expected that they will require lifelong support with daily living.

> ***Our son now lives in his own bungalow. We chose to use a direct payment which we use to employ his PAs who work with him 24/7. His PAs are like family now and we are confident he is well looked after and he is thriving."***

Unique publishes a separate guide to Transition

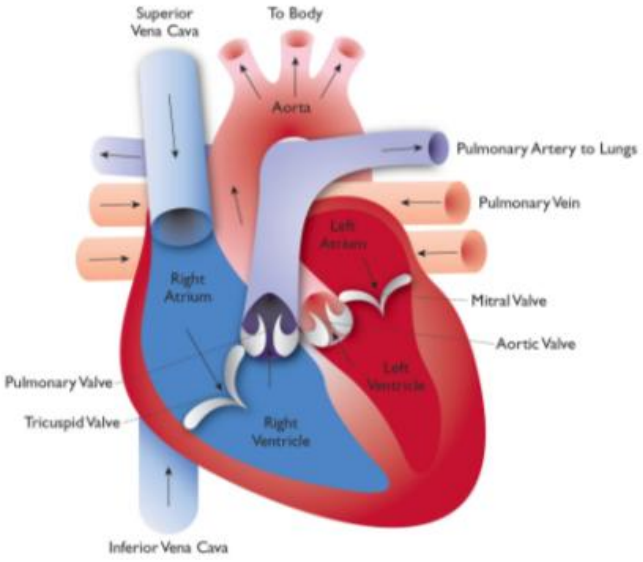


Medical concerns

The following medical concerns have been found in children with a 5p deletion. They are not found in all children so not all children with a 5p deletion will be affected.

Heart conditions

A heart condition(s) has been found in many people reported so far (2025) with a 5p deletion, which can be present at birth (congenital) or develop later in life. In children for whom heart problems are suspected, these can be diagnosed using tests like an electrocardiogram (ECG) (recording the electrical activity of the heart), echocardiogram (ultrasound scan of the heart), or chest X-ray. The type of heart condition(s) is variable but include anomalies affecting the size and structure of the heart muscle and valves. Most often these include:



A normal heart

- ASD (atrial septal defect - a hole between the top two chambers of the heart)
- VSD (ventricular septal defect - a hole between the bottom two chambers of the heart)
- PDA (persistent ductus arteriosus - failure of closure of the tube that carries blood between the aorta and the pulmonary artery during the foetal period)

Less commonly, there can be:

- Tetralogy of Fallot (a combination of four heart anomalies: VSD; pulmonary valve stenosis (narrowing of the pulmonary valve); right ventricular hypertrophy (thickening of the muscle of the right ventricle); and overriding aorta (the aorta isn't in its usual position coming out of the heart).
- Change in one of the heart valves (such as the pulmonary valve)
- Left ventricular outflow obstruction (restricted blood flow out of the left ventricle into the aorta)
- A double outlet right ventricle (both the aorta and the pulmonary artery are connected to the right ventricle)

Many of these conditions are relatively minor and resolve naturally in time. Medical treatment may be necessary for others, and some may require surgery.

Brain anomalies

Many people reported so far (2025) with a 5p deletion have a structural brain anomaly, which can be detected by MRI (magnetic resonance imaging) or a CT (computerised tomography) scan of their brain. The changes seen vary but include:

- A reduction in brain volume (cerebellar atrophy or pontine hypoplasia)
- Enlarged fluid-filled cavities in the brain (dilated ventricles)
- An underdevelopment (hypoplasia) or partial/complete absence (agenesis) of the white matter connecting the two halves of the brain (corpus callosum)

Less commonly, there can be:

- Areas of abnormal brain matter (focal cortical dysplasia)
- Dandy-Walker malformation (where an area at the back of the brain (cerebellum) that controls movement and balance does not develop properly)

Seizures

A few children with a 5p deletion experience some form of seizure (a sudden and unexpected change in the electrical activity in the brain). Depending on the part(s) of the brain affected, symptoms vary, but include temporary confusion, uncontrollable jerking movements and loss of consciousness or awareness. Age of onset can vary considerably, while seizures may be isolated to a single incident or occur more regularly. More than one type of seizure may be present in the same individual. Electroencephalograph (EEG) and video telemetry (video EEG) are medical tests that can be used to measure and record the electrical activity of the brain and are tools that, when used alongside other tests, can help diagnose the type of seizure experienced.

Seizures can cause a lot of worry for families and can be frightening to observe, but in some cases they self-resolve or resolve with medical treatment. If your child has a seizure for the first time, it is important to remove nearby hazards so they can't hurt themselves, and contact a medical professional.

Eyes and eyesight

Problems with eyes and vision are common in children with a 5p deletion. A wide range of conditions have been reported and an individual may have more than one vision or eye-related concern. Known concerns include:

- Short sightedness (myopia) which can usually be corrected by glasses
- Long-sightedness (hypermetropia) which can usually be corrected by glasses
- A slight alteration in eye shape that can lead to blurry vision (astigmatism)
- A squint (strabismus), where one eye or both turn inward, outward, up or down, which may be treated with patching, glasses, exercises or surgical correction
- A "lazy eye" (amblyopia), which can be a consequence of a constant squint in one eye
- Widely spaced eyes (hypertelorism)
- Eyes that are slightly downward slanted
- Retinal dystrophy (a range of chronic and progressive eye conditions affecting vision)

Hearing

Some children with a 5p deletion have a hearing impairment. Hearing is unaffected in other children and hearing tests at birth may give a clear response. A hearing loss may be conductive, where sound is unable to travel effectively to the inner ear; sensorineural, where there are problems with the inner ear, sometimes with the cochlea or auditory nerve (the nerve that sends signals to the brain about sound); or a combination of both conductive and sensorineural hearing loss.

Many types of hearing loss can be managed by using hearing aids. As children are at risk of speech delay, parental concerns should be acted on early and home- or school-based therapy provided.

Some children experience "glue ear", where fluid builds up behind the ear drum, which may be made worse by unusually narrow external ear canals and excess wax in the ear canal. Glue ear is a type of conductive hearing loss and is typically treated by inserting aeration tubes (grommets) into the eardrum. This surgical operation may need to be repeated. Improved hearing may not be achieved with aeration of the space behind

the eardrum (middle ear) and hearing aids may help as a temporary or longer-lasting measure, although this appears to be uncommon.

Unique publishes a separate guide to Hearing

Breathing

Babies and children with rare chromosome and gene disorders tend to have a higher rate of respiratory concerns, which may become less frequent with age and maturity, although they can persist throughout childhood. Children may also be prone to allergies and asthma, sometimes triggered by respiratory infections. Other issues may include snoring. Some parents describe their child's breathing as weak or noisy and this is often because the vocal cords are formed differently.

Hands and feet

Children with a 5p deletion occasionally have anomalies of the hands and feet. The most common among these include:

- Small hands and feet
- Fingers or toes that are unusually short (brachydactyly)
- Fingers or toes that are fused (syndactyly)
- Underdeveloped (hypoplastic) or misshapen nails
- Club foot (talipes) with the foot turned inwards and the soles pointing towards each other

A wide variety of other specific anomalies of toe and foot position are also an occasional feature. Some children are only mildly affected, and will not require treatment. Others may benefit from massage, orthotics and physiotherapy. Treatment is tailored to the individual child, and in some cases surgical correction will best enhance eventual mobility.

Spine

Many children with a 5p deletion are born with or develop a spinal curvature, most commonly a sideways curve of the spine (scoliosis). The curvature can be treated with physiotherapy and exercises, or a support brace or surgery may be needed. A sacral dimple (dimple or hole in the skin just above the crease between the buttocks) is also sometimes seen. The dimple may be shallow so you can see the base, but stools can collect there before your child is toilet-trained, so keeping it clean and protected is important. A sacral dimple may be deep and even connect to the spinal canal or the colon. If there is any concern about this, your baby's spine will be imaged, usually with ultrasound or an MRI scan.

Skin

Several children with a 5p deletion have a skin condition(s). Eczema, where the skin becomes red, itchy and inflamed can occur. Your doctor should be able to recommend self-care techniques, emollients and other treatments that may help to relieve symptoms.

Other skin conditions that have been reported include skin tags that either fall off naturally or can be surgically removed, and cutaneous haemangioma (an abnormal build-up of blood vessels on or under the surface of the skin that can look like a red-coloured birthmark).

Teeth

Dental concerns are very common in children with chromosome disorders. A number of issues have been described by parents of children with a 5p deletion including:

- Unusual dental development
- Unusual jaw size, leading to overcrowding or widely-spaced teeth
- Feeding difficulties and delayed eating and chewing activity
- Unusually thin, weak enamel (enamel hypoplasia)
- Tooth grinding (bruxism), which can prematurely wear down enamel
- Late emerging teeth
- Late loss of milk teeth

A high standard of dental care is important to minimise damage by decay and erosion (by grinding). Children and adults may also benefit from specialist hospital dental services and may require treatment under general anaesthetic.

Unique publishes separate guides to Looking after your child's teeth and Teeth: common concerns

Hernias

A few babies are born with a hernia, where an organ or fatty tissue pushes through a weak spot in a surrounding muscle or tissue. These include instances of:

- Umbilical (at or near the belly button)
- Inguinal (at or near the inner groin)

Less commonly, there can be:

- Abdominal (at or near the abdomen, between the chest and the pelvis)
- Diaphragmatic (in the diaphragm, the muscle that separates the chest cavity from the abdominal cavity)
- Hiatal (upper stomach)

In a few individuals the hernias healed naturally without the need for treatment, but in the majority of people, surgical repair was required.

Palate

Anomalies of the palate (roof of the mouth), ranging from those that may be invisible to the casual onlooker such as a high/arched palate to more obvious conditions such as a cleft palate, have been reported in children with a 5p deletion. Anomalies of the palate, particularly clefting, can cause difficulties in feeding, hearing, teething and speech production. As well as helping aesthetically, surgical repair eases these problems and may even eliminate them altogether.

Kidney and urinary tract

Occasionally babies are born with minor anomalies of the kidneys and/or urinary tract. Urinary tract infections (UTIs) sometimes occur and may need to be treated with antibiotics. Repeated urinary infections may require preventive treatment with antibiotics. Reported anomalies include:

- An enlarged kidney(s) (hydronephrosis) due to a build-up of urine inside, which may sometimes be diagnosed during mid-pregnancy anomaly scans. In mild cases this requires monitoring but no treatment. More serious cases can cause UTIs, which can be treated with antibiotics or, very occasionally, a catheter may need to be inserted to remove the build-up of urine and prevent damage to the kidney.
- Kidney (urethral) reflux, where urine flows upwards from the bladder back up to the kidney, potentially damaging the kidneys and leading to frequent UTIs.

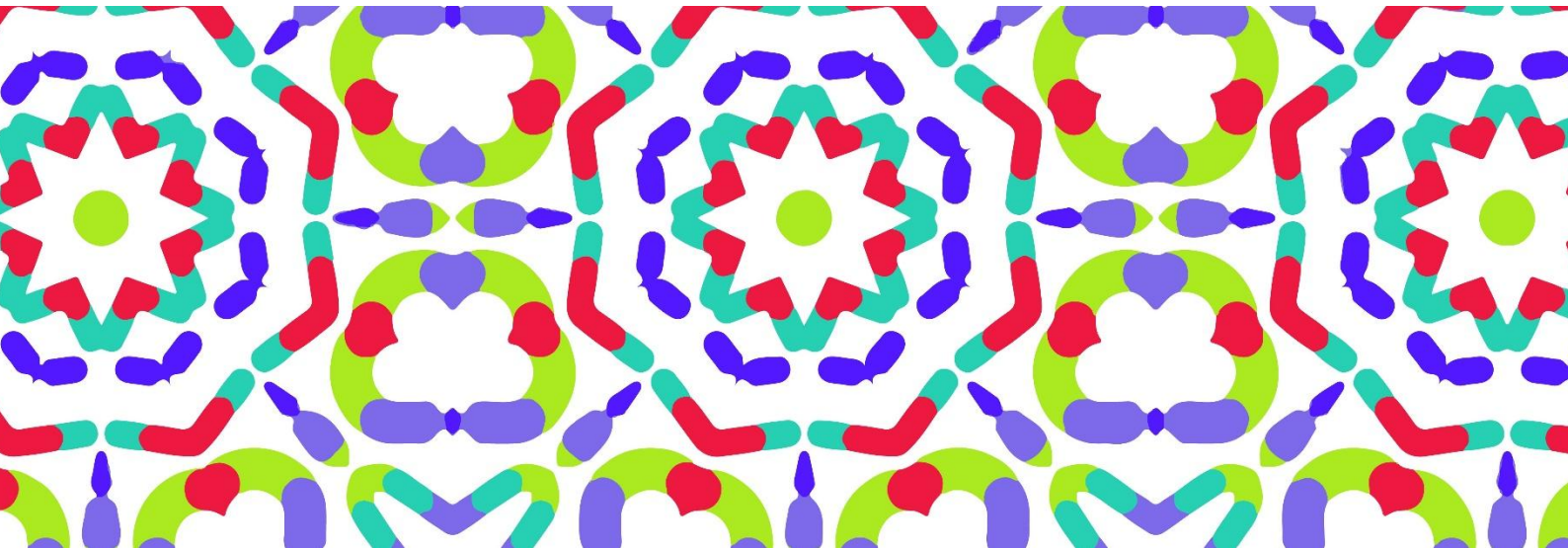
Less commonly, there can be:

- Having only one kidney
- Having a duplex kidney, where one or both kidneys have two ureter tubes to drain urine rather than a single tube, which can increase the risk of UTIs
- Benign renal caliectasis, where there is a slight ballooning of part of the kidney; or under-sized kidneys.

Genitals

Minor anomalies of the genitals in boys have been reported frequently. Most common among these are hypospadias, where the hole normally at the end of the penis lies on the underside; a curved penis; a very small penis (micropenis); a buried (hidden) penis, where the penis is partially or completely covered by skin; undescended testes (cryptorchidism). Many of these anomalies can also be seen in children without a 5p deletion and are not of major concern. If necessary, most can be corrected with surgery.

Girls are much less likely to be affected. Conditions that have been reported rarely include under-developed internal genitalia and labia.



How common is a 5p deletion?

A 5p deletion is extremely rare. Currently (2025) about 1 in 15,000 to 1 in 50,000 individuals are affected with a 5p deletion. However, among the autosomal chromosomal (not including the X or Y chromosomes) deletion disorders, 5p deletions are among the most common.

Why did this happen?

When children are conceived, their parents' chromosomes are copied in the egg and sperm that makes a new child. The biological copying method is not perfect, and random changes occur in the DNA of all children, that are not seen in the DNA of their parents. This happens naturally and is not due to the parents' diet, environment or lifestyle. Most of these DNA changes have no obvious effect. But in rare instances these random DNA changes can lead to health issues or affect development. In almost all people identified so far (2025) with a 5p deletion, the genetic change was a random (or "*de novo*") change, meaning the change occurred for the first time in that family in the affected individual. Very rarely, one parent may have the deletion themselves and passed it on to their child. This parent may be mildly or unaffected. Very rarely one parent has a chromosomal rearrangement that led to a 5p deletion in their child, or one parent may have the same deletion in some of their egg or sperm cells and pass it on to their child (this is known as germline mosaicism). However, it is important to recognize that no one should be blamed for their chromosome deletions and no parent is at fault when a new chromosome deletion occurs in their child.

Can it happen again?

The possibility of having another child affected by a rare gene disorder depends on the genetic code of the parents. In almost everyone reported with a 5p deletion so far (2025) the genetic change has been found to be *de novo* (dn), which means neither parent was found to have the same chromosome deletion as their child, and neither parent was found to have a chromosomal rearrangement that might have resulted in the deletion in their child. Therefore, the chance of having another child with a 5p deletion is usually less than 1%.

One reason why there is some residual chance of recurrence is due to the rare phenomenon called germline mosaicism that was mentioned above. This is when a parent carries a genetic change, but it is limited to some of their egg or sperm cells. The genetic change would not, therefore, be detected in the parents' blood tests.

Unique publishes a short general guide to Mosaicism

Parental genetic testing has been reported in the medical literature for several individuals with a 5p deletion and parental mosaicism for the deletion has only been reported once.

In rare families with a 5p deletion, a parent was found to be a carrier of a balanced chromosomal rearrangement. This is when a piece of chromosome is relocated, usually onto a different chromosome, but no DNA is lost. When a child is conceived, the change in the parent's chromosome(s) can lead to a deletion in one or more of the child's chromosomes. This parent therefore had a high probability of having further children with a 5p deletion. Again, this is very rare and has only been reported approximately 20 times so far (2025) in the medical literature.

Unique publishes a separate guide to balanced translocations

A clinical geneticist or genetic counsellor can provide specific advice for each family about the chance of having further children with a 5p deletion.

Unique publishes separate guides to Planning your next child, Prenatal genetic testing and diagnosis, A clinical genetics appointment and Supporting siblings of children with a rare genetic condition

Can a 5p deletion be cured?

There is no complete cure for a 5p deletion since most of the effects of the genetic change took place during a baby's formation and development. However, knowing the diagnosis means that appropriate monitoring and interventions can be put in place.

Management

Children and adults with a 5p deletion should be under the care of a multidisciplinary team. The team should include a geneticist and paediatrician (for children) who can oversee care so that development and behaviour can be monitored, and the best help given in the form of physiotherapy, occupational therapy, speech therapy and, if needed, behavioural therapy. Individuals may have evaluations with e.g. audiology, cardiology, endocrinology, gastroenterology, nephrology, neurology, ophthalmology, and urology.

Immediately following diagnosis

When not carried out as part of the diagnostic process, an evaluation of the features of a 5p deletion that are present in the child or adult who has been diagnosed with this genetic condition should be carried out. This can determine which of the features of a 5p deletion are present and how severe they are.

Supportive care

How a person with a 5p deletion is cared for is likely to require co-ordinated care by a multidisciplinary team of specialists, including a:

Paediatrician – a doctor who specialises in the physical, mental and social health of children from birth to young adulthood.

Cardiologist – a doctor who specialises in heart conditions.

Endocrinologist – a doctor who specialises in hormones and their effect on the body.

Nephrologist – a doctor who specialises in conditions affecting the kidneys.

Neurologist – a doctor who specialises in conditions of the brain, spinal cord and nervous system.

Ophthalmologist – a doctor who specialises in conditions affecting the eyes.

Surgeon - doctor who is specially trained to perform medical operations.

Urologist – a doctor who specialises in diagnosing and treating conditions affecting the urinary system.

Audiologist – a health care professional who diagnoses, treats and helps manage a condition that involves hearing or balance.

Occupational therapist (OT) – a health care professional who uses activities to aid self-management of a condition and can provide equipment.

Physiotherapist (PT) – a health care professional who uses exercise, movement, manual therapy, education and advice to help with the body's strength and mobility.

Speech and language therapist (SALT) – a health care professional who helps with speech, language communication and sometimes feeding/swallowing difficulties.

Psychiatrist – a doctor who specialises in mental health.

Specialist nurses and/or other healthcare professionals may need to systematically and comprehensively plan a child or adult's treatment.

Treatments and therapies

Treatment will depend on the specific features and symptoms experienced by the person with a 5p deletion but may include:

Physiotherapy for gross motor skills and/or low muscle tone which helps with strengthening of muscles and weight bearing.

Occupational therapy for fine motor skills which helps with feeding, hand use, coordination, and sensory issues.

Speech therapy for communication skills which helps with mouth movement, breath control, and sign language.

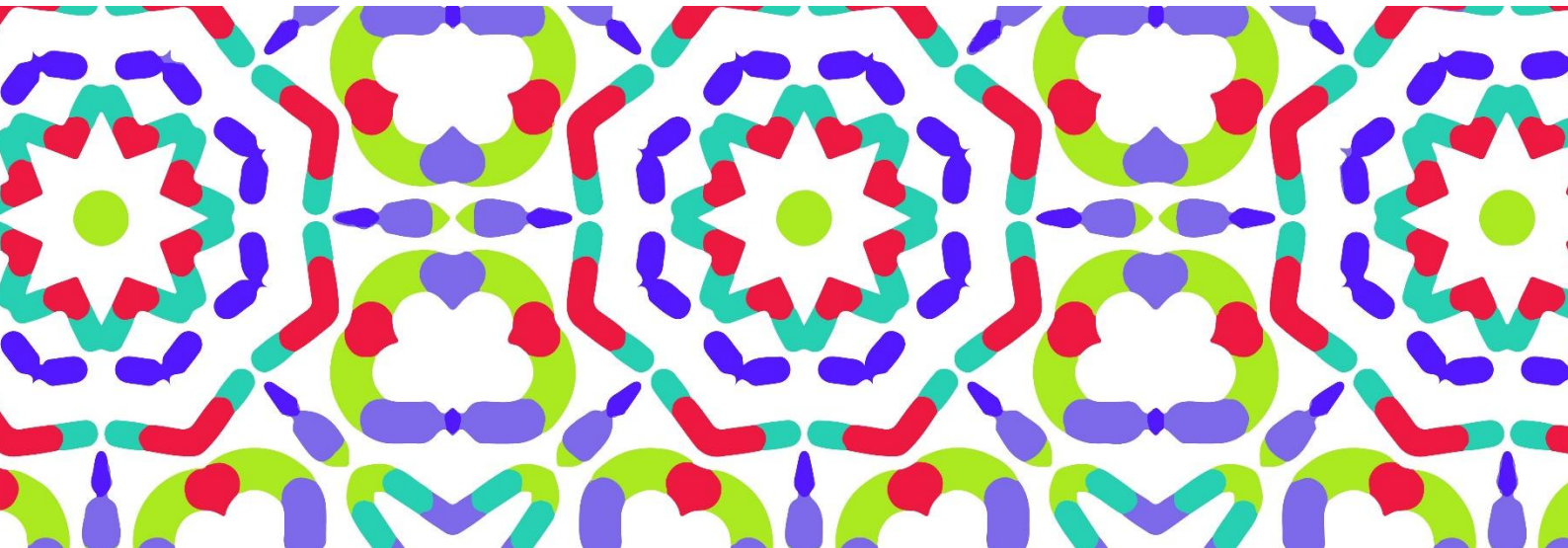
Behavioural therapy for sensory issues, sleep, autistic traits, anxiety, self-harming, aggressive behaviours, compulsive traits, inattentiveness, or hyperactive traits.

Medications may be prescribed depending on each person's specific symptoms.

Diet, such as a high-fibre diet or stool softeners and/or laxatives may be recommended to help relieve constipation. Some benefit from enemas when symptoms are particularly severe.

Surveillance

It is recommended that the following evaluations are carried out to monitor an individual's existing symptoms, how they respond to care and treatment, and whether any new symptoms emerge over time: growth, development, vision and hearing.



Is there any research into new treatments for a 5p deletion?

A 5p deletion affects development of the brain and other parts of the body before birth. Therefore, a complete cure is unlikely, even in the future, since the brain has already formed by the time a diagnosis is made. However, research into improved treatments and management for various features of chromosome deletions, like autism, is ongoing. In addition, although a 5p deletion is a relatively rare condition, the genes that have been deleted are often the subject of some research.

5p genes

This section provides further information about important genes included in 5p deletions. The entire p-arm of chromosome 5 contains about 300-400 genes and symptoms may depend on which genes are included in each child's specific deletion. A number of genes in this region are known to be expressed in the brain and have been associated with neurological function. There is currently limited information known about the relevance of the genes in this specific region. Further information about the p-arm of chromosome 5 as well as the genes found in this region of chromosome 5 is continuously gathered as researchers study the effects of genetic changes. Genes of interest may include:

CTNND2 - loss of one copy of this gene may be related to intellectual disability and neurodevelopmental deficits.

SEMA5A - loss of one copy of this gene may be associated with cognitive function as well as autistic features.

ICE1, UBE2QL1, or MARCH6 - loss of one copy of this gene may be implicated in the characteristic high-pitched cry.

TERT - loss of one copy of this gene may contribute to premature greying of hair.

TRIO – in people with larger deletions, loss of one copy of this gene this may be associated with developmental delay, intellectual disability, behavioural concerns, small head (microcephaly), feeding and growth concerns, constipation, finger or toe anomalies, scoliosis, dental abnormalities, seizures and heart anomalies.

There are occasional reports of rare cases where children have a gene deleted from 5p, and the same gene on their second chromosome 5 is altered in a different way, which means that there is not a copy of that particular gene that is working as it should be. It would therefore be advisable for a child with a 5p deletion who has more severe or uncommon symptoms to be reviewed (e.g. referred to clinical genetics) to consider whether further investigations are needed.

Families say ...

"Our son loves to socialise and has lots of friends. He enjoys meet-ups with friends, discos, bowling, games nights and lots more. Exercise is really important to maintain a range of movement so we do lots of walking and swimming etc."

Sources

The information in this guide is drawn from published medical literature, databases and Unique members. The first-named author and publication date from articles are given to allow you to look for the abstracts or free access articles on the internet in **PubMed**. Information gathered from **DECIPHER** (DatabasE of genomiC varIation and Phenotype in Humans using Ensembl Resources) is open access.

Unique currently (2025) has 65 members with Cri du Chat syndrome who live world-wide. Some members also have additional genetic changes, for these families, the reason for some of their symptoms and features may be due to additional genetic anomalies. This guide may however be of use to such families to partly explain their child's difficulties.

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Note: an asterisk indicates articles which are "open access" and available to everyone at **https://pubmed.ncbi.nlm.nih.gov**

Inform Network Support

Rare Chromosome Disorder Support Group
The Stables, Station Road West, Oxted, Surrey, RH8, 9EE, UK
Tel: +44(0)1883 723356
help@rarechromo.org I rarechromo.org

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Condition specific websites:

- **Five P Minus Society**

This guide was written and reviewed by The 5p Minus Society Professional Advisory Board, United States and Unique (AP).

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