Cri du chat syndrome
Summary

Cri du chat syndrome is a rare genetic disorder caused by missing pieces on a particular chromosome. It is not the result of anything the parents have done or failed to do. The characteristics of a newborn with cri du chat syndrome include a high-pitched cry, a small head and a flattened bridge of the nose. Treatment aims to help the child reach their full potential.

Cri du chat syndrome is a rare genetic disorder caused by a missing section on a particular chromosome known as Chromosome 5. Sometimes, material from another chromosome is missing as well. Around one in every 50,000 or so babies is diagnosed with this disorder. This estimate is coming down. With more advanced and frequent genetic testing, and many more milder cases being diagnosed, it may be as low as one in 25,000.

Around a third as many girls as boys are affected. Other names for this condition include cat-cry syndrome, 5P minus syndrome and Le Jeune’s syndrome.

Chromosomes are responsible for our inherited characteristics such as eye colour. In a child with cri du chat syndrome, some of the genes located on Chromosome 5 are missing, usually for reasons unknown. The impact of this can vary from one child to the next, but most will have delayed development and some degree of intellectual disability.

There is no cure, but treatments (including speech therapy, physiotherapy and occupational therapy) can help the child to reach their full potential.

Symptoms of cri du chat syndrome

Cri du chat syndrome is often diagnosed at birth. The symptoms in a newborn baby can include:

- a high-pitched, cat-like cry or weak cry
- low birth weight
- a small head
- a rounded face
- a broad, flattened bridge of the nose
- eyes spaced wide apart
- folds of skin over the eyelids
- abnormalities of the palate, such as an unusually narrow and high palate
- a receding, small chin
- malformations of the ears.

Health problems for children with cri du chat syndrome

Children with cri du chat syndrome commonly experience health problems including:

- difficulties with suckling and swallowing
- gastric reflux
- constipation
- high rate of infections, such as ear and upper respiratory tract infections
- poor muscle tone
- eye problems, such as a squint (strabismus).

Other problems, which occur less frequently, include:

- hearing loss
- deformities of the skeleton, such as curvature of the spine (scoliosis)
- hernias
- heart and kidney problems.

Causes of cri du chat syndrome

No one knows what causes the deletion of certain genes on chromosome 5. In most cases, the abnormality happens spontaneously, with no family history. The risk of a couple with normal chromosomes having another child with cri du chat is around one per cent. In some cases, one of the parents has abnormalities of Chromosome 5.

Genetic counselling for these couples is important, as subsequent children may also have cri du chat syndrome. Medical techniques such as amniocentesis, where a sample of amniotic fluid is examined, can determine whether or not an unborn baby has cri du chat syndrome. Amniocentesis is usually performed in the first trimester of pregnancy.
Severity of cri du chat syndrome varies

The severity of the condition can vary. Some children are only mildly affected, and reach their developmental milestones, such as walking and talking, at the usual ages. However, they may still need speech therapy. Other children are profoundly intellectually disabled, unable to walk or talk, and suffer from related health problems and a reduced life span.

The majority of children born with cri du chat syndrome fall between these two extremes. They have moderate intellectual disability, with varying degrees of speech delay and some health problems, but most master walking some time before the age of six. It is impossible to predict whether or not a newborn baby with cri du chat syndrome will be mildly, moderately or severely affected. This will only become clear as the child gets older.

Treatment for cri du chat syndrome

There is no cure for cri du chat syndrome. Treatment aims to stimulate the child and help them to reach their full potential and can include:

- physiotherapy to improve poor muscle tone
- speech therapy
- communication alternatives, such as sign language, since speech is usually delayed, often severely
- occupational therapy to teach coping strategies and new skills.

Support for parents of children with cri du chat syndrome

Support services for parents of children with disabilities include:

- counselling
- information
- referral
- advocacy
- support groups.

Where to get help

- Your doctor
- Cri du chat Support Group of Australia Tel. (03) 9775 9962
- Association for Children with a Disability Tel. (03) 9880 7000 or 1800 654 013
- Better Start for Children with a Disability Tel. 1800 242 636

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More information

Genes and genetics

The following content is displayed as Tabs. Once you have activated a link navigate to the end of the list to view its associated content. The activated link is defined as Active Tab

- A-Z of genetic conditions
- Genes and genetics explained
- Genetic testing

A-Z of genetic conditions

- Ambiguous genitalia
  
  The causes of ambiguous genitalia include genetic variations, hormonal imbalances and malformations of the fetal tissues that are supposed to evolve into genitals.
- Angelman syndrome
  
  The characteristic features of Angelman syndrome are not always obvious at birth, but develop during childhood.
- Ankylosing spondylitis
  
  Ankylosing spondylitis (AS) is a type of inflammatory arthritis that targets the joints of the spine.
- Barrett's oesophagus
  
  Symptoms of Barrett's oesophagus are similar to regular heartburn, which means many people don't seek treatment until their condition is quite advanced.
- Bipolar disorder
  
  Bipolar disorder is a type of psychosis, which means the person's perception of reality is altered. It is characterised by extreme mood swings.
- Central nervous system birth defects
  
  Folic acid taken before conception, and during at least the first four weeks of pregnancy, can prevent around seven out of 10 cases of neural tube defects.

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- Charcot-Marie-Tooth disease (CMT)
  Charcot-Marie-Tooth disease is the most common inherited disorder affecting the peripheral nervous system.

- Cleft palate and cleft lip
  Most cleft palates and cleft lips can be repaired so that appearance and speech develop normally.

- Congenital adrenal hyperplasia (CAH)
  CAH is a rare genetic disorder, but it is well understood and treatment is readily available.

- Creutzfeldt-Jakob disease (CJD)
  Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, dementia and walking difficulties.

- Cri du chat syndrome
  Most children born with cri du chat syndrome have moderate intellectual disability, with varying degrees of speech delay and some health problems.

- Cystic fibrosis (CF)
  When a person has cystic fibrosis, their mucus glands secrete very thick sticky mucus that clogs the tiny air passages in the lungs and traps bacteria.

- Digestive tract birth defects
  Too much amniotic fluid surrounding the baby during pregnancy (polyhydramnios) may indicate the presence of defects of the digestive tract.

- Down syndrome
  With the support and opportunities available to them today, most people with Down syndrome are able to achieve and participate as valued members of their community.

- Dwarfism
  Dwarfism refers to a group of conditions characterised by shorter than normal skeletal growth.

- Eczema (atopic dermatitis)
  Eczema can vary in severity, and symptoms may flare up or subside from day to day.

- Essential tremor
  Essential tremor causes involuntary shaking or trembling of particular parts of the body, usually the head and hands, but it is not Parkinson's disease.

- Fragile X syndrome
  The facts about fragile X syndrome are complicated, and parents and family members are invited to ask their doctor to refer them to a genetics clinic.

- Friedreich's ataxia
  To the casual observer, a person with Friedreich ataxia may seem to be drunk.

- Genetic factors and cholesterol
  Familial hypercholesterolaemia is an inherited condition characterised by higher than normal levels of blood cholesterol.

- Haemochromatosis
  Haemochromatosis (iron overload disorder) tends to be under-diagnosed, partly because its symptoms are similar to those caused by a range of other illnesses.

- Haemophilia
  All children with severe haemophilia are given preventative treatment with infusions of blood products before they have a bleed.

- Hair
  Human hair grows one centimetre every month.

- Hearing problems in children
  The earlier that hearing loss is identified in children, the better for the child's language, learning and overall development.

- Huntington's disease
  The symptoms of Huntington's disease usually, but not always, first appear when the person is approaching middle age.

- Kabuki syndrome
  Kabuki syndrome affects males and females equally and there is no cure.

- Kennedy's disease
Kennedy's disease is a rare inherited neuromuscular disorder that causes progressive weakening and wasting of the muscles, particularly the arms and legs.

- Kidneys - medullary cystic kidney disease
  Medullary cystic kidney disease causes the growth of abnormal cysts in the kidneys.

- Kidneys - polycystic kidney disease (PKD)
  Polycystic kidney disease is a common cause of kidney failure in Australia and equally affects men and women of different ethnic backgrounds.

- Klinefelter syndrome
  Klinefelter syndrome is often diagnosed at puberty, when the expected physical changes don't occur.

- Leukodystrophy
  Leukodystrophy refers to a group of inherited disorders that affect the white matter of the brain, which causes loss of normal brain functions.

- Long QT syndrome
  You should be investigated for long QT syndrome if you faint for no apparent reason, during or after exercise or emotional excitement.

- Marfan syndrome
  Some people may not realise they have Marfan syndrome, because their features are either mild or not obvious.

- McCune-Albright syndrome
  The severity of symptoms or how a child with McCune-Albright syndrome will be affected throughout life is difficult to predict.

- Muscular dystrophy
  People affected by muscular dystrophy have different degrees of independence, mobility and carer needs.

- Neurofibromatosis
  Neurofibromatosis is caused by faulty genes, which may be inherited or have spontaneously mutated at conception.

- Noonan syndrome
  Noonan syndrome is a genetic condition that usually includes heart abnormalities and characteristic facial features.

- Osteoporosis in children
  Osteoporosis in children is rare and usually caused by an underlying medical condition.

- Phenylketonuria (PKU)
  PKU is an inherited disorder that prevents the normal breakdown of a protein found in some foods.

- Prader-Willi syndrome
  A feature of Prader-Willi syndrome is the child's excessive appetite, which often leads to obesity.

- Premature and early menopause
  The symptoms of premature or early menopause are the same as for menopause at any age.

- Rett syndrome
  People with Rett syndrome have a keen desire to communicate.

- Spinal muscular atrophy (SMA)
  A child with spinal muscular atrophy type 1 rarely lives beyond three years of age.

- Tay-Sachs disease
  Tay-Sachs disease is a serious genetic disorder common in Ashkenazi Jews and French-Canadians.

- Thalassaemia
  Thalassaemia is an inherited blood disorder that can cause anaemia or death if not treated.

- Tourette syndrome
  Milder forms of Tourette syndrome can be misdiagnosed, as it often occurs at the same time as attention deficit hyperactivity disorder (ADHD) and other disorders.

- Treacher Collins syndrome
  Treacher Collins syndrome is a genetic disorder that affects growth and development of the head, causing facial defects and hearing loss.
• Tuberous sclerosis
  Tuberous sclerosis is a genetic disorder that affects various parts of the body to varying degrees of severity...

• Turner's syndrome
  Turner's syndrome is a random genetic disorder that affects females, causing short stature and infertility...

• Usher syndrome
  Services aim to help a person with Usher syndrome prepare for the dual loss of sight and hearing...

• Von Willebrand disease
  A person with von Willebrand disease may have frequent nosebleeds, heavy menstruation or excessive bleeding from the mouth...

• Williams syndrome
  Williams syndrome often goes undiagnosed, which means that some people with the disorder fail to get the support and treatment they need until later in life...

• Wilson disease
  In Wilson's disease, a build-up of copper damages organs including the liver, nervous system, brain, kidneys and eyes...

Genes and genetics explained

• Genes and genetics explained
  Children inherit physical characteristics such as eye colour from their parents through their genes...

• Gene therapy
  Gene therapy is an experimental form of treatment that targets the faulty genes that cause genetic diseases...

• Genetic disorders
  genetic disorder is caused by an altered or faulty gene or set of genes...

• Genetic services in Victoria
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare...

Genetic testing

• DNA profiling
  DNA profiling is a way of establishing identity and is used in a variety of ways...

• Egg freezing
  You can freeze your eggs for medical reasons or for reasons that are more to do with your life circumstances...

• Genetic services in Victoria
  Genetic services can help people who are affected by, or who are at risk of, inherited conditions or birth defects, to make informed choices about their healthcare...

• Genetic testing for inherited cancer
  A predisposition to certain cancers can be inherited via altered genes...

• Newborn bloodspot screening
  Every newborn baby in Australia is offered a newborn bloodspot screening test to identify those at risk of rare, but serious, medical conditions...

• Pregnancy tests - chorionic villus sampling
  Chorionic villus sampling (CVS) is a pregnancy test that checks the baby for some abnormalities...

• Pregnancy tests - maternal serum screening
  Maternal serum screening can indicate increased risk of abnormalities in the unborn child, but is not a diagnosis...

Related Information

• Down syndrome
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**Related information on other websites**

- Centre for Developmental Disability Health (CDDHV)
- Cri Du Chat Support Group of Australia brochure (218 kb, pdf)
- genetic support network victoria
- MedlinePlus Medical Encyclopedia: Cri du chat syndrome
- Spine Society of Australia - National Self-Detection Program for Scoliosis

**Support Groups**

- Cri Du Chat Support Group of Australia

**Content Partner**

This page has been produced in consultation with and approved by: Cri du Chat Support Group of Australia

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Cri du Chat Syndrome

This condition is also called cri-du-chat or 'cry of the cat' syndrome. It is a genetic condition that affects the ability of newborns to cry. This syndrome is also known as del(47,XX,-15,+,t(47,XX,-15,15q21.3)).

## What is cri-du-chat syndrome?

Cri-du-chat syndrome is a genetic disorder. It is a rare genetic condition that occurs in newborns and is characterized by the inability to cry. This condition affects newborns and is often diagnosed soon after birth. The genetic cause of this syndrome is a deletion of the 5th chromosome and it is often lethal in the first year of life.

### Symptoms of cri-du-chat syndrome

The main symptoms of cri-du-chat syndrome include:

- **Inability to cry at birth**: This is the main symptom of the syndrome and is often what leads to the diagnosis.
- **Frequent respiratory infections**: Due to the immune system being weakened.
- **Low body weight**: Usually below the 10th percentile.
- **Low head circumference**: Often below the 10th percentile.
- **Seizures and other neurological problems**: These can occur in some cases.
- **Heart problems**: These can occur in some cases.
- **Gastrointestinal (GI) problems**: These can occur in some cases.
- **Cataracts**: These can occur in some cases.
- **Haematuria (blood in the urine)**: This can occur in some cases.
- **Developmental delays**: These can occur in some cases.

### Diagnosis of cri-du-chat syndrome

Diagnosis of cri-du-chat syndrome is made through a combination of symptoms and genetic testing. The genetic test involves a chromosome analysis, which can confirm the presence of the deletion.

### Causes of cri-du-chat syndrome

Cri-du-chat syndrome is caused by a deletion of a portion of chromosome 5. The deletion occurs in the region 5p15.3-p15.2.

### Treatment of cri-du-chat syndrome

Treatment of cri-du-chat syndrome is focused on managing the symptoms and complications. This can include:

- **Supportive care**: This includes helping the child meet their developmental needs.
- **Medical interventions**: These may include medications for seizures, infections, and other medical conditions.
- **Special education and therapies**: These may include physical therapy, occupational therapy, and speech therapy.
- **Ongoing monitoring**: This includes regular check-ups to monitor the child's development and health.

### Support for cri-du-chat syndrome

Support for cri-du-chat syndrome includes:

- **Family support**: This includes emotional support and practical assistance.
- **Community support**: This includes support groups and other community resources.
- **Professional support**: This includes the support of healthcare professionals and specialists.

### Prevention of cri-du-chat syndrome

Cri-du-chat syndrome is not preventable. However, genetic counseling can help parents understand the risks of having a child with this condition.

### Conclusion

Cri-du-chat syndrome is a rare genetic condition that affects newborns. It is characterized by the inability to cry at birth and can be diagnosed through a combination of symptoms and genetic testing. Treatment is focused on managing the symptoms and complications and providing ongoing support for the child and their family.

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