Cri du chat syndrome
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 sucking 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

Send us your feedback

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- Your details

1/4 How would you rate this website?

- Excellent
- Good
- Average
- Fair
- Poor

Please note that we cannot answer personal medical queries. If you are looking for health or medical advice we recommend that you:

- talk to your doctor or pharmacist
- dial triple zero (000) in an emergency
- ring NURSE-ON-CALL Tel. 1300 60 60 24.

2/4 Your Comments

Tell us who you are Select an option
- **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.

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.

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- **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

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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]

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### Content Partner

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

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