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## Cri du chat syndrome

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### 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.
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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.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by cri du chat syndrome.

### **Genetic counselling and cri du chat syndrome**

If your child has been diagnosed with cri du chat syndrome, or if it runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand the condition and what causes it, how it is inherited, and what a diagnosis means for your child's health and for your family. Genetic counsellors are trained to provide information

and support that is sensitive to your family circumstances, culture and beliefs.

If cri du chat syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

#### **Where to get help**

- **Your GP (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**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**

**This page has been produced in consultation with and approved by:**

Cri du Chat Support Group of Australia

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