

Cri du chat syndrome

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, however, coming down and 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

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

- High pitched, cat-like cry or weak cry
- Low birth weight
- Small head
- Rounded face
- 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
- Receding, small chin
- Malformations of the ears.

Health problems

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.

The cause is unknown

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.

The degree of severity

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 sometime 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. Only time will tell.

Treatment options

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.

Help for the parents

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) 9561 8134 or phone/fax (03) 9775 9962
- Association for Children with a Disability Tel. (03) 9500 1232 or 1800 654 013

Things to remember

- 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.

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

Cri du Chat Support Group of Australia

Copyright © 1999/2009 State of Victoria. Reproduced from the Better Health Channel (www.betterhealth.vic.gov.au) at no cost with permission of the Victorian Minister for Health. Unauthorised reproduction and other uses comprised in the copyright are prohibited without permission.

- This Better Health Channel fact sheet has passed through a rigorous approval process. For the latest updates and more information visit www.betterhealth.vic.gov.au.

