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Cri du chat syndrome

Cri du chat syndrome is a group of symptoms that result from missing a piece of chromosome number 5. The syndrome's name is based on the infant's cry, which is high-pitched and sounds like a cat.

Causes

Cri du chat syndrome is rare. It is caused by a missing piece of chromosome 5.

Most cases are believed to occur during the development of the egg or sperm. A small number of cases occur when a parent passes a different, rearranged form of the chromosome to their child.

Symptoms

Symptoms include:

- Cry that is high-pitched and may sound like a cat
- Downward slant to the eyes
- Epicanthal folds, an extra fold of skin over the inner corner of the eye
- Low birth weight and slow growth
- Low-set or abnormally shaped ears
- Hearing loss
- Heart defects
- Intellectual disability
- Partial webbing or fusing of fingers or toes
- Curvature of the spine (scoliosis)
- Single line in the palm of the hand
- Skin tags just in front of the ear
- Slow or incomplete development of motor skills
- Small head (microcephaly)
- Small jaw (micrognathia)
- Wide-set eyes

Exams and Tests

Your health care provider will perform a physical exam. This may show:

- Inguinal hernia
- Diastasis recti (separation of the muscles in the belly area)
- Low muscle tone
- Characteristic facial features

Genetic tests can show a missing part of chromosome 5. Skull x-ray may reveal any problems with the shape of the base of the skull.

Treatment

There is no specific treatment. Your provider will suggest ways to treat or manage the symptoms.

Parents of a child with this syndrome should have genetic counseling and testing to determine if one parent has a change in chromosome 5.

Support Groups

More information and support for people with Cri du chat syndrome and their families can be found at:

- 5P- Society -- fivepminus.org [https://fivepminus.org/]

Outlook (Prognosis)

Intellectual disability is common. One half of children with this syndrome learn enough verbal skills to communicate. The cat-like cry becomes less noticeable over time.

Possible Complications

Complications depend on the amount of intellectual disability and physical problems. Symptoms may affect the person's ability to care for themselves.

When to Contact a Medical Professional

This syndrome is most often diagnosed at birth. Your provider will discuss your baby's symptoms with you. It is important to continue regular visits with the child's providers after leaving the hospital.

Genetic counseling and testing is recommended for all people with a family history of this syndrome.

Prevention

There is no known prevention. Couples with a family history of this syndrome who wish to become pregnant may consider genetic counseling.

Alternative Names

Chromosome 5p deletion syndrome; 5p minus syndrome; Cat cry syndrome

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Updated by: Anna C. Edens Hurst, MD, MS, Associate Professor in Medical Genetics, The University of Alabama at Birmingham, Birmingham, AL. Review provided by VeriMed Healthcare Network. Also reviewed by David C. Dugdale, MD, Medical Director, Brenda Conaway, Editorial Director, and the A.D.A.M. Editorial team.