

Causes, Symptoms and Diagnosis of Cri Du Chat Syndrome

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DESCRIPTION

Cri du chat syndrome, also known as 5p-syndrome, is a rare genetic disorder caused by the deletion or loss of a portion of chromosome 5. The name cri du chat means cry of the cat in French, referring to the distinctive high-pitched cry that affected infants produce, which sounds similar to the meowing of a cat.

Causes

Cri du chat syndrome is primarily caused by a deletion of genetic material on the short arm of chromosome 5. It typically happens during the formation of reproductive cells (eggs or sperm) or in early embryonic development. The exact cause of these random deletions is unknown. However, in rare cases, Cri du chat syndrome can be inherited from a parent who carries a balanced translocation involving chromosome 5. In a balanced translocation, a rearrangement of genetic material occurs, but there is no overall loss or gain of genetic material. When a parent with a balanced translocation involving chromosome 5 passes on the translocated chromosome to their child, it can result in Cri du chat syndrome.

Symptoms

The symptoms and severity of Cri du chat syndrome can vary widely among affected individuals. Common features include the high-pitched cry in infancy, intellectual disability, delayed development, distinctive facial features (such as a small head, round face, wide-set eyes, and a small jaw), low birth weight, weak muscle tone (hypotonia) and microcephaly (abnormally small head size). Other possible symptoms include feeding difficulties, speech and language delays, behavioral problems and medical issues like heart defects and vision or hearing problems.

Diagnosis

The diagnostic process involves:

Clinical evaluation: A healthcare professional, such as a pediatrician or geneticist, will conduct a thorough evaluation of the individual's medical history, as well as assess their physical

and developmental characteristics. The distinctive cry of affected infants, resembling the meowing of a cat, is often one of the first signs noticed.

Physical examination: The healthcare provider will examine the individual for physical features associated with Cri du chat syndrome. These may include a small head (microcephaly), a round face, wide-set eyes, a small jaw, low-set ears, and other distinctive facial characteristics. They will also assess for developmental delays, muscle tone and any other abnormalities.

Genetic testing: To confirm the diagnosis, genetic testing is usually performed. This can involve different techniques.

A karyotype analysis examines the chromosomes to identify any abnormalities, including a deletion on the short arm of chromosome 5. This test can help determine if the individual has Cri du chat syndrome.

Fluorescent *In Situ* Hybridization (FISH) is a more specific test that uses fluorescent probes to detect and visualize the missing portion of chromosome 5. It can provide a precise confirmation of the 5p deletion.

Array comparative genomic hybridization test can identify small deletions or duplications of genetic material and may be used to detect other chromosomal abnormalities or genetic syndromes that can coexist with Cri du chat syndrome.

Prognosis

Some key factors to consider regarding the prognosis:

Intellectual disability: The majority of individuals with Cri du chat syndrome experience some level of intellectual disability. The severity can range from mild to profound. Early intervention programs, educational support, and therapies can help individuals with Cri du chat syndrome reach their full potential and acquire functional skills.

Developmental delays: Delayed development is common in individuals with Cri du chat syndrome. Milestones such as sitting, walking, and speech may be achieved later than expected. Early intervention services, including physical therapy, occupational therapy, and speech therapy, can assist in

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addressing developmental delays and promoting progress in various areas.

Language and communication: Speech and language development can be significantly affected in Cri du chat syndrome. Some individuals may develop functional language skills, while others may have limited verbal communication abilities. Alternative communication methods such as sign language or Augmentative and Alternative Communication (AAC) systems can be helpful.

Medical issues: Cri du chat syndrome is associated with various medical problems that can affect the overall health and well-being of individuals. These may include heart defects, respiratory issues, gastrointestinal problems, hearing loss, and vision problems. Regular medical monitoring and appropriate

interventions can help manage these issues and improve outcomes.

Behavioral and psychological challenges: Individuals with Cri du chat syndrome may exhibit behavioral challenges, such as attention deficits, hyperactivity, impulsivity, and self-injurious behaviors. Behavioral interventions, counseling, and support from mental health professionals can assist in managing these challenges and improving overall behavior and quality of life.

Lifespan: In general, individuals with Cri du chat syndrome have a normal lifespan. However, it's important to note that the associated health issues, particularly heart defects and respiratory problems, can impact longevity. Regular medical care and proactive management of medical conditions are crucial for maintaining good health.