Cri Du Chat Syndrome

What is Cri du chat Syndrome?

Cri du Chat Syndrome arises from the absence of a particular portion of chromosome 5. The absence of genetic material is called a deletion. Children born with this deletion have a characteristic mewing cry in that is thought to be caused by abnormal development of the larynx (the organ containing the vocal chords). For this reason, the disorder is called Cri du Chat (or Cat’s Cry) Syndrome. In reference to the deletion and the area where it occurs, the disorder is also called 5p- (5 p minus) Syndrome. Children with this syndrome have physical abnormalities, language and motor skill difficulties, and varying degrees of intellectual disabilities. Cri du Chat Syndrome is a very rare disorder.

Who is affected?

The estimated prevalence is about 1 in 50,000 live births

How is it manifested?

• high-pitched cry (identified as a cat-like cry)
• low birth weight
• poor muscle tone (hypotonia)
• microcephaly (small head size)
• micrognathia (small jaw)
• hypertelorism (wide spaced eyes)
• round face
• epicanthal folds (small skin folds on the inner corners of the eyes)
• low set ears
• feeding difficulties
• delay in walking
• hyperactivity
• scoliosis
• language difficulties
• intellectual disabilities
• organ defects

How is it diagnosed or detected?

During infancy, a diagnosis of Cri du Chat Syndrome is strongly suspected if the characteristic cat-like cry is heard. If a child has this unusual cry, or other features seen in Cri du Chat Syndrome, chromosome testing should be performed. Chromosome analysis provides a definitive diagnosis of Cri du Chat Syndrome and can be performed from a blood test. Chromosome analysis, also called “karyotyping,” involves staining the chromosomes and examining them under a microscope. In some cases the deletion of material from Chromosome 5 can be easily seen. In other cases, further testing must be performed. FISH (Fluorescence In-Situ Hybridization) is a special technique that detects very small deletions. The majority of the deletions that cause Cri du Chat Syndrome can be identified using the FISH technique.

Additional Resources:

5P- SOCIETY - www.fivepminus.org
The 5p- Society (five p minus), a not-for-profit corporation, is the parent support group for families having a child with 5p- Syndrome, also known as Cat Cry Syndrome or Cri du Chat Syndrome. Their mission is to encourage and facilitate communication among families having a child with 5p- Syndrome and to spread awareness and education of the syndrome to these families and their service providers.
ALLIANCE OF GENETIC SUPPORT GROUPS
- www.geneticalliance.org
Genetic Alliance is an international coalition comprised of more than 600 advocacy, research and healthcare organizations that represent millions of individuals with genetic conditions and their interests. As a broad-based coalition of key stakeholders, they leverage the voices of millions of individuals living with genetic conditions.

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