Cri-du-chat syndrome

Description

Cri-du-chat (cat's cry) syndrome, also known as 5p- (5p minus) syndrome, is a chromosomal condition that results when a piece of chromosome 5 is missing. Infants with this condition often have a high-pitched cry that sounds like that of a cat. The disorder is characterized by intellectual disability and delayed development, small head size (microcephaly), low birth weight, and weak muscle tone (hypotonia) in infancy. Affected individuals also have distinctive facial features, including widely set eyes (hypertelorism), low-set ears, a small jaw, and a rounded face. Some children with cri-du-chat syndrome are born with a heart defect.

Frequency

Cri-du-chat syndrome occurs in an estimated 1 in 20,000 to 50,000 newborns. This condition is found in people of all ethnic backgrounds.

Causes

Cri-du-chat syndrome is caused by a deletion of the end of the short (p) arm of chromosome 5. This chromosomal change is written as 5p-. The size of the deletion varies among affected individuals; studies suggest that larger deletions tend to result in more severe intellectual disability and developmental delay than smaller deletions.

The signs and symptoms of cri-du-chat syndrome are probably related to the loss of multiple genes on the short arm of chromosome 5. Researchers believe that the loss of a specific gene, *CTNND2*, is associated with severe intellectual disability in some people with this condition. They are working to determine how the loss of other genes in this region contributes to the characteristic features of cri-du-chat syndrome.

Learn more about the gene and chromosome associated with Cri-du-chat syndrome

- CTNND2
- chromosome 5

Inheritance

Most cases of cri-du-chat syndrome are not inherited. The deletion occurs most often as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. Affected people typically have no history of the disorder in their family.

About 10 percent of people with cri-du-chat syndrome inherit the chromosome abnormality from an unaffected parent. In these cases, the parent carries a chromosomal rearrangement called a balanced translocation, in which no genetic material is gained or lost. Balanced translocations usually do not cause any health problems; however, they can become unbalanced as they are passed to the next generation. Children who inherit an unbalanced translocation can have a chromosomal rearrangement with extra or missing genetic material. Individuals with cri-du-chat syndrome who inherit an unbalanced translocation are missing genetic material from the short arm of chromosome 5, which results in the intellectual disability and health problems characteristic of this disorder.

Other Names for This Condition

- 5p deletion syndrome
- 5p- syndrome
- Cat cry syndrome
- Chromosome 5p- syndrome
- Monosomy 5p

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: 5p partial monosomy syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0010314/)

Genetic and Rare Diseases Information Center

- Monosomy 5p (https://rarediseases.info.nih.gov/diseases/6213/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Cri-du-chat syndrome

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Catalog of Genes and Diseases from OMIM

- CRI-DU-CHAT SYNDROME (https://omim.org/entry/123450)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Cri-du-Chat+Syndrome%5BMAJR%5D%29+AND+%28%28cri-du-chat+syndrome%5BTIAB%5D%29+OR+%285p-%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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Last updated October 25, 2022