

Cri Du Chat

Cri Du Chat is the result of a congenital partial deletion that most commonly occurs de novo. Like most other microdeletions, this one causes a syndrome.



PLAY PICMONIC

Pathophysiology

Congenital Deletion of Short Arm of Chromosome 5p

Cutting off Chromosome of (5) Hand on Short Arm

Cri Du Chat occurs as a result of a de novo partial deletion of Chromosome 5p. As such, it is classified as one of the "5p- syndromes".

Signs and Symptoms

High Pitched Cry/Mewing

High Pitched Crying Meow

A hallmark of Cri Du Chat is a high-pitched cry due to an underdeveloped, dysmorphic larynx. The high-pitched cry usually resolves by age 2.

Microcephaly

Small-head

Microcephaly, or a shrunken head, is the most prominent clinical feature of Cri Du Chat syndrome. Research has shown a strong genotype-phenotype correlation between the degree of severity of the microcephaly and the amount of partial deletion that occurs in 5p.

Hypotonia

Floppy Hippo-baby

Patients with Cri Du Chat will present with hypotonia, failure to thrive, and developmental delay.

Difficulty Sucking and Swallowing

Difficulty Sucking

Due to microcephaly and psychomotor retardation, patients with Cri Du Chat have difficulty with sucking and swallowing.

Epicanthal Folds

Epicanthal Folds

Epicanthal folds occur when the skin of the upper eyelid covers the inner corners of the eyes, a common feature seen in Cri Du Chat.

Widely Spaced Eyes

Wide Spaced Eyes

Patients typically have orbital hypertelorism, resulting in an abnormally wide space in between the patient's eyes.

Wide & Flat Nasal Bridge

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Another common feature in patients with Cri Du Chat is a wide and flat nasal bridge.



High Arched Palate

High Arched Palette

A high-arched palate is a variable feature in patients with Cri Du Chat. The palate is narrow and high.

Intellectual Disability

Tar Covered Book

The severity of the intellectual disability is dependent on the size of the deletion.

Cardiac Abnormalities

Abnormal Heart

Congenital heart defects (most commonly VSD), pneumonia, and respiratory distress syndrome are the most common causes of death in patients with Cri Du Chat.