

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

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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

[Wide & Flat Nasal Bridge](#)

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.