

A Note on 5p Minus Syndrome

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DESCRIPTION

Cri-du-chat syndrome could be a genetic condition. Additionally called Cat cry syndrome, 5p minus syndrome and chromosome 5p deletion syndrome. This disease is under the class of congenital and genetic diseases. It's a rare disorder, arising in newborns. "Cri-du-chat" means "cry of the cat" in French. Infants with this condition typically have a high-pitched cat-like cry. The larynx organ develops abnormally because of the chromosome deletion that affects the sound of the child's cry.

SYMPTOMS

The severity of child's symptoms depends on what quantity genetic information is missing from chromosome five. Some symptoms are severe whereas others are so minor they may go unknowingly. The cat-like cry is the most common symptom becomes less noticeable over time. The subsequent list includes the most common signs and symptoms.

- Babies with cri-du-chat are usually tiny at birth, and will have metabolism issues.
- Often, the larynx organ doesn't develop properly, that causes the cat like cry.
- Mental retardation.
- Small head (microcephaly).
- Small jaw (micrognathia).
- Downward slant of the eyes.
- Wide-set eyes.
- Abnormally shaped/positioned ears.
- Webbed fingers or toes.
- Single line within the palm of the hand (simian crease).
- Hanging skin before of the ears.
- Slow or incomplete development of basic skills.

CAUSES

Cri du chat syndrome is activated by a missing piece (deletion) of the short (p) arm of chromosome five. This chromosomal modification is known as (5p-). The size and site of the deletion varies among affected people.

- Cri-du-chat is initiated by a deletion on the small arm of chromosome 5p.
- The measurement of the deletion might vary.
- Several genes are missing as a result of this deletion, and each may contribute to the symptoms of this condition.
- HTERT (human telomerase reverse transcriptase) helps to keep the information in DNA functioning properly. If HTERT is broken, mental diseases like cri-du-chat occur.

Inheritance

Most cases of cri-du-chat syndrome don't seem to be hereditary. The deletion happens most frequently as a random event during the formation of reproductive cells (eggs or sperm) or in early foetal development. Affected individuals usually don't have any history of the disorder in their family. People with cri du chat syndrome inherit unbalanced translocation with genetic material omitted from the short arm of chromosome five.

DIAGNOSIS

Doctors most frequently determine cri-du-chat by looking at the patient's symptoms. Another methodology of identification cri-du-chat syndrome takes place while the baby was in its mother's uterus. Doctors will either check a small sample of tissue from outside the sac where the baby develops, or they will check a sample of the liquid chromosome substance.

TREATMENT

Treatment for cri du chat syndrome is focused on the managing the symptoms. Specialists who can be concerned within the care of somebody with cri du chat syndrome include:

- Medical geneticist
- Physical therapist
- Occupational therapist
- Speech therapist
- Ear, nose, and throat (ENT) specialist
- Behavior or development specialist

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PREVENTION

There are no known ways to prevent the cri-du-chat syndrome. Even if an individual doesn't show symptoms, he is also a carrier

as that individual includes a case history of the syndrome of his family. Cri-du-chat syndrome is extremely rare, thus it's unlikely to have more than one kid with the condition.