Original Research Paper

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CRI- DU- CHAT SYNDROME (CAT'S CRY) ALSO KNOWN AS LEJEUNE'S SYNDROME, ASSOCIATED WITH LEARNING DISABILITIES, ANATOMICAL ABNORMALITIES INCLUDING GASTRO INTESTINAL AND CARDIOVASCULAR PROBLEMS

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ABSTRACT Cri du chat syndrome, also known as 5p- (5p minus) syndrome or cat cry syndrome, is a genetic condition that is caused by the absence of genetic material on the small arm (the p arm) of chromosome 5. The absence of 5p occurs most often during the of reproductive cells (eggs or sperm) formation or in early fetal development. Common manifestations are learning disabilities, anatomical abnormalities including GI and cardiovascular problems. Children may be treated by speech, physical and occupational therapists. Heart abnormalities often require surgical correction. There is no cure for cri du chat syndrome.

KEYWORDS : Cri-du-chat, 5p-, Chromosome, Fetal Development.

INTRODUCTION

Cri du chat syndrome (CdCS or 5p-) is a rare genetic disorder in which a variable portion of the short arm of chromosome 5 is missing or deleted. In 1963 the disorder was first described by doctor Lejeune who observed abnormal cat-like cry in newborn. In French, Cri du chat means "cry of the cat".Cri du chat syndrome(cat's cry). 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 peculier 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.

DEFINITION

Cri du chat syndrome, also known as 5p- (5p minus) syndrome or cat cry syndrome, is a genetic condition that is caused by the absence of genetic material on the small arm (the p arm) of chromosome 5.

INCIDENCE

Cri-du-chat syndrome occurs in an estimated 1 in 20,000 to 50,000 newborns. This condition is more in people of all ethnic backgrounds. The condition affects an estimated 1 in 50,000 live births across all ethnicities and is common in females by a 4:3 ratio.

CAUSES

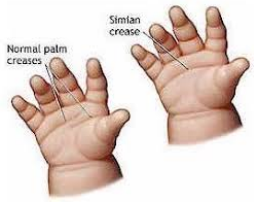
- 90% of the cases are not inherited. The absence occurs most often as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. Most affected individuals do not have a history of the disorder in their family.
- Caused by a absence of the end of the short (p) arm of chromosome 5. This chromosomal change is written as 5p-
- 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.
- Cri du chat syndrome - also known as 5p- syndrome and cat cry syndrome - is a rare genetic condition that is causedby the absence (a missing piece) of genetic material on the small arm (the p arm) of chromosome 5. The cause of this rare chromosomal absence is unknown.
- The loss of a specific gene, CTNND2

SIGNS AND SYMPTOMS

High pitch cry resambles to meowing kitten, due to problems with the larynx and nervous system. It can subside at the age of 2 years.

Other symptoms of cri du chat syndrome may include:

- feeding problems due to difficulty in swallowing and sucking.
- low birth weight and poor growth.
- severe cognitive, speech and motor disabilities.
- behavioral problems like hyperactivity, aggression, outbursts and repetitive movements;
- unusual facial features, which may change over time.
- excessive drooling of saliva.
- microcephaly, micrognathism and hypertelorism.
- Skin tags in front of eyes.
- Other common findings include hypotonia, round face with full cheeks, epicanthal folds, down-slanting palpebral fissures (eyelids), squint, flat nasal bridge, down-turned mouth, low-set ears, short fingers, single palmar creases and cardiac defects (e.g. tetralogy of Fallot).



- difficulties in communicating, especially child feels difficulty in complete the sentence.
- cleft lip and palate, preauricular tags and fistulas, thymic dysplasia, intestinal malrotation, megacolon, cryptorchidism, hypospadias, dislocated hips, inguinal hernia, , rare renal malformations (e.g., horseshoe kidneys, renal ectopia or agenesis, hydronephrosis), various dermatoglyphics, including transverse flexion creases, distal axial triradius, increased whorls and arches on digits and a single palmar crease also includes clinodactyly of the fifth fingers, talipes equinovarus, pes planus, syndactyly of the second and third fingers and toes, oligosyndactyly and hyperextensible joints are rare in this condition.
- Significant findings include intellectual disability, microcephaly, coarsening of facial features, prominent supraorbital ridges, deep-set eyes, hypoplastic nasal bridge, severe malocclusion and scoliosis in adolescence.
- Affected females reach puberty, develop secondary sex characteristics and menstruate at the usual time. The genital tract is usually normal in females, except for a report of a bicornuate uterus. In males, testes are often small, but spermatogenesis is thought to be normal.
- Exceptionally, some with Cri du chat are very high-functioning as developmentally typical individuals, with mostly the exception of mild learning difficulties, and do not have speech difficulties, although they may have milder facial features and a high-pitched voice due to their condition.

DIAGNOSIS

- Cri du Chat syndrome can be diagnosed clinically, by physical characteristics.
- A genetic test involves taking a sample of blood or saliva to detect absence on chromosome 5.
- Antenatally-diagnosed cri-du-chat syndrome, Although PAPP-A (Pregnancy Associated Plasma Protein A) was low at first trimester screening (FTS), the combined risks of trisomies 21, 18 and 13 were low.
- Amniocentesis carried out following the ultrasonographical observation of a severely hypoplastic nasal bone, cerebellar hypoplasia, choroid plexus cyst and a single umbilical artery during the second trimester.
- FISH analysis (fluorescent in situ hybridization). If there is a

family history of cri-du-chat, suggest a chromosome analysis or genetic testing while child is still in the womb.
- Chorionic villus sampling or test a sample of amniotic fluid with BACs-on-Beads (Bacterial Artificial Chromosomes) technology, G-banded karyotype of a carrier is also useful for detecting missing of chromosome.
- Genetic testing registry: Genetic counseling and genetic testing may be offered to families with individuals who have cri du chat syndrome.

TREATMENT

There is no specific treatment for cri-du-chat syndrome. Manage symptoms with physical therapy, language and motor skill therapy, and educational intervention.

There is no known way to prevent cri-du-chat syndrome where as prenatal screening can helps to detect early diagnosis.

Heart abnormalities often require surgical correction.

Treatment aims to stimulate the child and help them to reach their full potential include:

- physiotherapy to improve poor muscle tone and the areas of physical therapy (achieving physical and motor milestones such as sitting and standing up), communication (speech therapy, sign language instruction), behavioral modification (for hyperactivity, short attention span, aggression), and learning (special education).
- speech therapy
- communication alternatives, such as sign language, since speech is usually delayed, often severely

Occupational therapy to teach coping strategies and new skills.
Support services for parents of children with disabilities include:

- counseling
- information
- referral
- advocacy
- Support groups.

PROGNOSIS

There is no cure for cri du chat syndrome. Most individuals with cri du chat syndrome have a normal life expectancy. A small number of children with this condition are born with serious organ defects and other life-threatening medical problems. These children may have a worse prognosis.

CONCLUSION

However, most individuals with Cri-du-chat syndrome live into adulthood. There are adults with Cri-du-chat syndrome in the medical literature who have been reported to live over the age of fifty with better treatments. Both children and adults with this syndrome are usually friendly and happy, and enjoy social interaction. Early and consistent educational intervention, along with physical and language therapy children who have 5p- Syndrome are capable of reaching their fullest potential and can lead full and meaningful lives.

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