Cri du chat Syndrome: A Case Report

Tommy Vu, DDS, Rodney Vergotine BChD, MSc

University of Michigan School of Dentistry / Mott Children's Health Center/Hurley Medical Center

Abstract

Cri du chat Syndrome is a genetic disorder characterized by distinctive craniofacial features and severe cognitive and psychomotor delay. A thirteen-year-old patient visited the University of Michigan Pediatric Dentistry clinic to establish dental care. The patient has a history of cerebral palsy, cognitive and developmental delay, small intestines, and difficulty swallowing. Extraoral features revealed distinctive facial appearance of wide nasal bridge, short philtrum, and hypertelorism. Intraoral findings included mandibular retrognathism, anterior open bite, and macroglossia. The dental management of this patient will be discussed in this case report, while providing common clinical features of individuals with Cri du chat syndrome.

Background

Cri du chat syndrome, also known as 5-p deletion syndrome, is a rare autosomal genetic disorder. Originally described by Jerome Lejeune in 1963, cri du chat is the French phrase for "cry of the cat", which is a distinctive characteristic of individuals with this syndrome, most commonly during infancy.

It is found in more females than males (3:1), having an incidence rate of one in 37,000 individuals. It does not have any geographical or ethnic associations.

Etiology: Cri du chat is caused by a total or partial deletion of the short arm of the 5-p chromosome. Within the 5-p chromosome, the deletion of the 5p15.3 is more commonly associated with the catlike cry characteristic, while the 5p15.2 is associated with dysmorphism and mental retardation.

Diagnosis: Testing is often completed while considering the maternal age or abnormal ultrasound findings. Conventional cytogenetics and Comparative Genomic Hybridization, in addition to fetal DNA testing through the maternal blood, are ways to test and diagnose for Cri du chat. Diagnosis is typically made within the first year of life.

Clinical Features

Cri du chat translates to "cry of the cat." Though not present in every individual, this can be used as a distinctive early indicator of the condition. It has been described as a monotonal weak catlike cry at birth. This is often due to laryngeal or epiglottic abnormalities.

Common craniofacial features may include:

-Wide nasal bridge
-Epicanthus
-Flattened maxilla
-Facial asymmetry
-Short neck
-Labial incompetence
-Short philtrum
-Low set ears
-Hypertelorism
-Divergent strabismus
-Thin and short upper lip

Common dental features may include:

-Mandibular retrognathism
-Class II malocclusion
-Macrodontia, macrostomy
-Hypotonic perioral muscle
-Dental anomalies (opacities, agenesis, supernumerary, hypoplasia)
-Anterior open bite
-High palate
-Macroglossia

Cardiac conditions: Though the incidence of cardiac anomalies are undetermined, a complete cardiac evaluation is encouraged. Some studies estimated 15-25% have cardiac anomalies, with the risk increasing to 55% based on the genetic translocation. Ventricular/atrial septal defects, patent ductus arteriosus, Tetralogy of Fallot, and other complex cardiac diagnoses have been found.

Other characteristics that may be present:

-Intellectual disability, language delay, learning disabilities, delayed psychomotor development
-Swallowing and feeding problems: This can lead to low birth weight, developmental delay, and risk for aspiration and pneumonia
-Microcephaly: The most common brain anomaly, however, hypoplasia and agenesis of the corpus callosum has been recorded

Clinical Management

- Family involvement is encouraged in educational and rehabilitation programs to understand and facilitate psychomotor, linguistic, and relationship development
- The clinical features and symptoms of individuals with Cri du chat can vary from mild to severe, a multidisciplinary approach for evaluation and treatment is encouraged. This includes orthodontists, ophthalmologists, cardiologists, neurologists, gastroenterologists, and otolaryngologists
- Applied intervention for behavior management is advised. Individuals with Cri du chat may have severe behavior problems including self-injurious, aggressive, or stereotyped-manner behaviors. However, they typically have sociable and cheerful personalities.
- If patients are undergoing general anesthesia, special attention is required by anesthesiologists due to possible airway difficulties from laryngeal or epiglottis abnormalities. Having a laryngeal mask ready is encouraged in case intubation is failing
- Language delay may be evident; however, individuals have shown to understand more complex language than what is expressed. Understanding their level of receptive language can aid in behavior management

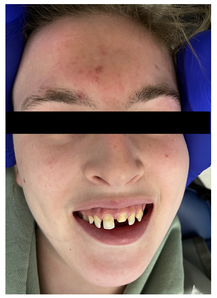
Dental management:

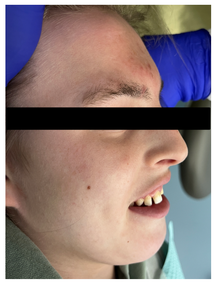
- Excellent oral hygiene instruction with 6 month recalls is encouraged as calculus formation is common in at least one quadrant with possible poor oral hygiene
- Due to multiple factors that may be contributing to the patient's behavior, treatment under general anesthesia may be indicated (90.2% in one study)
- Dental erosive lesions caused by gastrointestinal reflux are possible, as well as enamel hypoplasia, opacities, and agenesis of teeth
- Anticipatory guidance with emphasis on dental trauma is encouraged, especially if self-injurious behavior is present

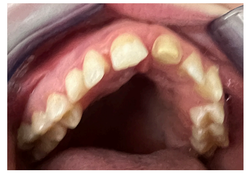
Case Report

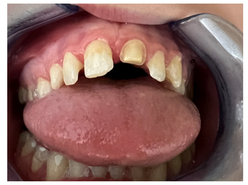
Patient MK:

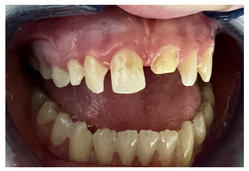
- 13- year, one- month old female presented to the pediatric dental clinic at the University of Michigan
- Medical History: Cri du chat syndrome (5-p deletion), cerebral palsy, cognitive and developmental delay, dysphagia
- Medications: none
- Allergies: NKA
- Family History: Lives at home with Mom and Dad
- Dental history: Sees mobile dentist at school 2x/year
  - Referred from a pediatric health center for evaluation and treatment
  - Chief complaint: "has cavities, missing crown"

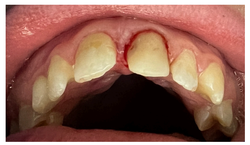












Extraoral examination: Facial presentation consistent with Cri du chat syndrome: wide nasal bridge, hypertelorism, short philtrum
Intraoral examination: Permanent dentition, high arched palate, Class II malocclusion, generalized moderate gingivitis
Exam findings: Caries detected clinically on #14-O and #19-O, missing restoration on #9

Caries risk assessment: high caries risk due to previous caries history and presence of caries
Behavior: Condition appropriate, minimal movements, tries to be cooperative, friendly with everyone
Treatment plan: Composite restorations on #9-MIDFL, #14-O, and #19-O with protective stabilization
Outcomes assessment: High risk fracture rate for #9-MIDFL, waiting until patient is older for full coverage crown, complete treatment plan, followed by 6 month recall for hygiene appointments

Conclusion:

Cri du chat Syndrome is a genetic disorder characterized by distinctive craniofacial features, cognitive and developmental delays, and systemic considerations. There are dental considerations including high risk for caries and periodontal concerns due to poor oral hygiene and malocclusion. In office dental treatment will depend on the level of treatment and child's behavior. Treatment under general anesthesia may be indicated.

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