

Oral manifestations of a patient with cri du chat (5p-) syndrome

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ABSTRACT

Cri du chat syndrome (CdCs) is a rare genetic disorder caused by deletion of the end of the short arm of chromosome 5. Affected infants are characterized by a high-pitched monochromatic cry that sounds similar to that of a meowing kitten. The incidence ranges from 1:15,000 to 1:50,000 live-born infants. Oral features of this condition include micrognathia, malocclusions, high but rarely cleft palate, anterior open bite, poor oral hygiene, enamel hypoplasia, chronic periodontitis, and retardation of tooth eruption. This case report presents typical medical and dental symptoms and dental rehabilitation of a 2.5-year-old female with CdCs. The patient was referred to our clinic because of caries arising from poor oral hygiene. The dental symptoms of the patient included hyperemic gingival tissue, abundant plaque, enamel hypoplasia, and tooth decay. Restorative treatment of the caries was completed and oral hygiene recommendations and caries risk factors were presented to the parents.

Key words: 5p Deletion, Cri Du Chat Syndrome, Orofacial Features, Pediatric Dentistry

Access this article online

Website:

www.jpediatrdent.org

DOI:

10.4103/2321-6646.155572

Quick Response Code:



INTRODUCTION

The cri du chat syndrome (CdCs) is a genetic disease^[1] in which affected infants are characterized by a cat-like cry.^[2] It was described by Lejeune in 1963, and the estimated frequency is between 1:15,000 and 1:50,000 live-born infants.^[3] The overall incidence of CdCs is generally higher among female births.^[2]

The combination of fluorescence *in situ* hybridization, comparative genome hybridization, and cytogenetic analysis revealed that CdCs is associated with a deletion in the short arm of 5p.^[4]

The clinical features include severe mental retardation; microcephaly; moon face; low-set ears; broad nasal bridge; abnormal dermatoglyphics; downward-slanting palpebral fissures; cardiac, neurological, and renal abnormalities; high-pitched monochromatic cry; hypertelorism; strabismus; and laryngeal abnormalities.^[5-8]

The main orofacial anomalies reported include mandibular microretrognathia, dental biprotrusion, dental malocclusions,

high but rarely cleft palate, anterior openbite, poor oral hygiene, enamel hypoplasia, generalized chronic periodontitis, and retardation of tooth eruption.^[3,6,9,10,11]

In this paper, we describe cranio-dento-facial findings and dental treatment in a 2.5-year-old female CdCs patient. Preventive dentistry is an important issue for pediatric dentists and the present case report emphasizes caries risk factors associated with CdCs.

CASE REPORT

A 2.5-year-old female was referred to the Department of Pediatric Dentistry at Akdeniz University for untreated cavities, difficulty swallowing, poor feeding, and poor oral hygiene. The parents reported that the patient had been diagnosed with CdCs. She was the second child of a healthy nonconsanguineous marriage and was born in the 36th week of gestation with a birth weight of 2600 g. The mother had taken antibiotics in the third trimester because of an urinary tract infection. The prenatal examination results were normal, and the karyotypes of the patient's sister and parents were normal. The

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medical history of the patient's family does not include any mental retardation nor consanguineous marriage. As the patient does not have a history of seizure, we did not consider magnetic resonance imaging or other tests to be necessary.

Written informed consent was obtained from the patient's parents for publication of this case report and identifiable images.

The patient weighed 9 kg (<3 centile) and her height was 85 cm (10 centile). She exhibited poor feeding, difficulty swallowing, and a cat-like cry. Her dysmorphic features included microcephaly, broad forehead, depressed nasal bridge, downward-slanting palpebral fissures, synophrys, bilateral low-set ears, high arched palate, anterior open bite, and simian lines on both hands [Figures 1-3]. An ophthalmologic examination revealed strabismus. An audiological examination revealed conductive hearing loss. Abdominal ultrasonography and echocardiography both showed normal results. Routine biochemistry test results were all within normal limits. Conventional cytogenetic tests revealed that the patient's karyotype was 46, XX, del (5p) (p15.1),13pss.

Intraoral findings included caries in all first primary molars, smooth-surface caries and enamel hypoplasia in anterior

teeth, together with abundant plaque and hyperemic gingivae [Figures 4 and 5]. The patient's parents revealed that because of her swallowing difficulty related with the syndrome she could only have liquid or soft foods. There was no toothache. In addition, there were no clinical symptoms indicating pulp inflammation. We didn't take dental radiographs because the patient was 2.5-year-old.

It was observed that a thumb-sucking habit had worsened the degree of dental malocclusion. Dental treatment was performed with the mother holding the patient in her arms. To maintain patient cooperation, an atraumatic restorative treatment technique, without any use of electrically driven equipment, was employed. Carious lesions were removed using hand excavators. Then, caries lesions, which were observed not to involve the pulp, were restored with high-viscosity glass-ionomer cement (EQUIA, GC, Tokyo, Japan) [Figure 6]. Fluorid varnish (Duraphat, Colgate Palmolive, New York, NY, USA) was applied to the children. Casein phosphopeptide-amorphous calcium phosphate paste (Tooth Mousse, GC, Tokyo, Japan) was recommended to the parents for twice a day.



Figure 1: The patient at 1-year of age



Figure 3: Single horizontal lines on the patient's hands



Figure 2: Facial view of the patient



Figure 4: View of the oral cavity

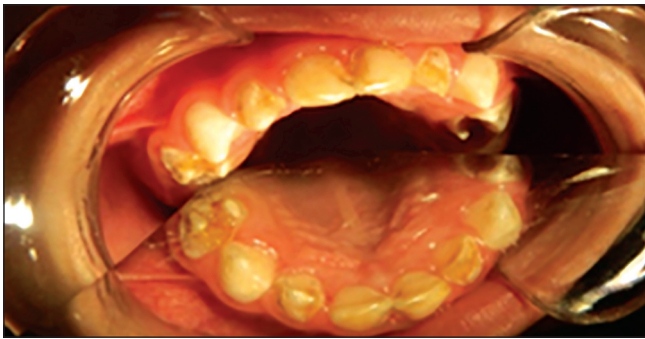


Figure 5: Dental caries and enamel hypoplasia

The parents were informed about risk factors for dental caries due to her feeding habits and lack of physiological cleaning. Techniques to improve the child's oral hygiene are provided to the parents.

DISCUSSION

Cri du chat syndrome (5p-syndrome) is a rare genetic syndrome exhibits hyperactivity and low intellectual age^[12] together with dental and cranial abnormalities that interfere with maintenance of adequate oral hygiene. This report describing a patient with CdCs provides information about typical features, particularly dental aspects, and treatment of dental manifestations.

A recent study including 15 patients with CdCs reported various congenital heart defects such as atrial septal defect (53%), ventricular septal defect (26%), tricuspid regurgitation (26%), and patent ductus arteriosus (20%), and laryngomalacia as the most frequent airway problem.^[13] However, in the present case, there was no congenital heart disease.

Besides systemic manifestations, orofacial findings were also reported. Although a previous study found that *Tannerella forsythia*-a periodontal pathogenic bacterial species-was more prevalent in the CdCs patient group than in the control group, no patients were found to suffer from active periodontitis.^[14] The present patient did not have chronic periodontitis either; however, there was abundant plaque and gingivitis because of poor oral hygiene.

The present patient exhibits enamel hypoplasia which is a caries risk factor and smooth-surface caries. Intrauterine enamel formation may be affected by fetal disorders or low-birth weight infants. When there is a genetic and/or enviromental condition at birth or in the neonatal period, corresponding enamel defects may be observed in the primary dentition. Developmental abnormalities including early feeding problems (due to swallowing difficulties and poor suck) and failure to thrive cause hypomineralized

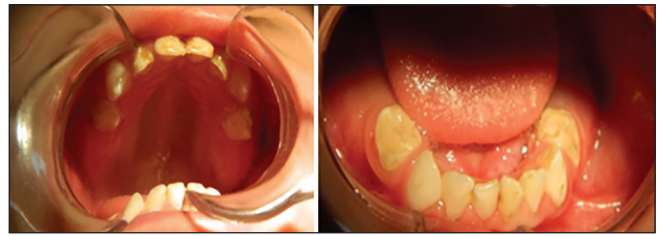


Figure 6: After the dental treatment

of teeth. Not only childhood factors but also prenatal lifestyle factors are important in this situation.^[15]

Intubation difficulties linked to larynx and epiglottis malformations are risk factors that may cause anaesthesiological problems.^[16] But, at an older age, many patients underwent general anaesthesia without complications.^[7,17] We decided to treat our patient with physical restraint at clinic. In order to stabilize the oral situation and control the number of pathogen microorganism in the oral cavity by release of fluorid, we decided to restore the present caries lesions with glass-ionomer cement. The glass-ionomer cement was also the most appropriate material because of noncooperation of the patient. Besides restorations, methods that prevent dental caries are applied. The use of fluoride has been a major factor in the decline in the prevalence and severity of dental caries. Although meticulous oral hygiene, and appropriate dietary practices contribute to caries prevention and control, the most effective and widely used approaches have included fluoride use. Fluoride varnish, by coating varnish on the surface of deciduous teeth, helps to increase the concentration of fluoride in enamel.

The choice of material for restoration to use in a given situation, however, is not simple. Amalgam is of limited use as further breakdown can often occur at the margins; and as it is nonadhesive, it does not restore the strength of the tooth. Stainless steel crown result in much higher success rates than multi-surface filling with any material. The use of glass-ionomers in the managment of decays in posterior primary teeth may be an excellent method in a 2.5-year-old child due to cooperation problems of the patient. By the age of 5 or 6 years, the child may be able to cope with more definitive treatment with composite resin, strip crown and stainless steel crowns.^[18]

Studies have shown that CdCs patients have skeletal Class 2 malocclusion due to mandibular retrognathia, dental biprotrusion, and small upper airway.^[3] Our patient had anterior open bite and mandibular growth retardation. These findings are similar to those of Breg *et al.*, who reported protrusion of the anterior teeth, dental malocclusion, openbite, and short metacarpals.^[19] Orthodontic management should be addressed with a long term comprehensive plan. This must involve several

dental disciplines such as orthodontics, periodontics, pediatric dentistry working together to coordinate a treatment plan. However, the patient is too young for the cephalometric analysis or radiograph. An orthodontist should consider interceptive orthodontic measures that reduce the degree of malocclusion in the mixed dentition stage. The patient will be able to manage conventional orthodontics with appropriate level of support.^[18] Retarded tooth eruption has been reported in the literature,^[3] but the tooth eruption in the patient in this case was within normal time.

The cranial base, cerebellum, and larynx are involved in CdCs, and a new theory involving a cranial developmental field comprising the dorsum sellae, clivus, cerebellum, and larynx has been suggested.^[20] Our patient had microcephaly and laryngeal developmental problems, and the presence of laryngomalacia explains the malnutrition and the cat-like cry. Cornish ve Pigram reported that 96.2% of individuals with CdCs can feed themselves.^[23] However, there is not any report about nature of foods. The parents of our patient informed that they are feeding their girl by only liquid or soft foods. The patients with CdCs suffer dental erosions provoked by gastroesophageal reflux.^[21] These were other risk factors of caries.

Behavioral problems, hyperactivity, lack of attention, self-injury, and aggressive behaviors have been reported.^[22,23] Our patient was too young at the time of the examination to identify any behavioral problems or attention deficit disorder, and the parents did not report any self-injury or aggressive behavior. Nevertheless, studies have shown that language and social-interaction skills can be developed in affected individuals.^[23]

CONCLUSION

it is important to recognize the orofacial and dental features of CdCs. This condition is associated with poor oral hygiene, high risk of caries, and periodontal tissue problems; therefore, personalized oral health plans and regular routine follow-up are advisable. Pediatric dentists can help to improve the dental health of affected individuals and inform their families about risk factors together with caries prevention and management methods.

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How to cite this article: Köylüoğlu ZI, Yıldız E, Nur BG, Mihçi E, Clark ÖA. Oral manifestations of a patient with cri du chat (5p-) syndrome. *J Pediatr Dent* 2015;3:67-70.

Source of Support: Nil. **Conflict of Interest:** None declared.