Reporte de casos
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Oral manifestations in child patient with Ellis-van Creveld syndrome: case report

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Abstract: Ellis–van Creveld syndrome is an autosomal recessive disorder characterized by a tetrad of disproportionate dwarfism, ectodermal dysplasia, postaxial polydactyly, and congenital heart malformations. In this article, we hereby present a case of a 6-year-old Brazilian boy with Ellis–van Creveld syndrome who presented with a remarkable number of classical oral and dental features and uncommon findings such as taurodontism. Clinical examination revealed multiple enamel hypoplasia, absent vestibular sulcus, alveolar serrations in the maxilla anterior region, missing teeth, conical teeth, lower canine rotation, bilateral posterior crossbite, dental caries, and a nodule. Radiographically were observed teeth agenesis, taurodontism of deciduous and permanent molars, and delayed tooth eruption. Clinical and radiographic findings may be present from birth and the pediatric dentist has a fundamental role in the early diagnosis of Ellis–van Creveld syndrome, as well as oral problems prevention, rehabilitation, and aesthetic interventions.

Key words: congenital heart disease, dental caries, pediatric dentistry, Ellis-van Creveld syndrome.

Manifestaciones orales en paciente infantil con síndrome de Ellis-van Creveld: reporte de caso

Resumen: El síndrome de Ellis-van Creveld es un trastorno autosómico recesivo caracterizado por una tétrada de enanismo desproporcionado, displasia ectodérmica, polidactilia postaxial y malformaciones cardíacas congénitas. En este artículo, presentamos el caso de un niño brasileño de 6 años con síndrome de Ellis-van Creveld que presentó un número notable de características orales y dentales clásicas y hallazgos poco comunes como taurodontismo. El examen clínico reveló hipoplasia múltiple del esmalte, surco vestibular ausente, aserraduras alveolares en la región anterior del maxilar, dientes ausentes, dientes cónicos, canino inferior girado, mordida cruzada posterior bilateral, caries dental y un nódulo. Radiográficamente se observa agenesia dentaria, taurodontismo de molares primarios y permanentes y retraso en la erupción dentaria. Los hallazgos clínicos y radiográficos pueden estar presentes desde el nacimiento y el odontopediatra tiene un papel fundamental en el diagnóstico precoz del síndrome de Ellis-van Creveld, así como en la prevención de problemas orales, rehabilitación e intervenciones estéticas.

Palabras clave: cardiopatías congénitas, caries dental, odontología pediátrica, Síndrome de Ellis-van Creveld.

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Manifestações orais em paciente infantil com síndrome de Ellis-van Creveld: relato de caso

Resumo: A síndrome de Ellis-van Creveld é uma doença autossômica recessiva caracterizada por uma tétrade de baixa estatura desproporcional, displasia ectodérmica, polidactilia pós-axial e malformações cardíacas congênitas. Neste artigo, será relatado um caso de um menino brasileiro de 6 anos de idade com síndrome de Ellis-van Creveld que apresenta um número notável de características orais e dentárias clássicas e achados incomuns como taurodontismo. Ao exame clínico foi revelado hipoplasia múltipla do esmalte, sulco vestibular ausente, serrilhas alveolares na maxila anterior, dentes ausentes, dentes cônicos, canino inferior rotacionado, mordida cruzada posterior bilateral, cárie dentária e um nódulo. Radiograficamente, foi observado agenesia dentária, taurodontismo de molares decíduos e permanentes e atraso na erupção dentária. Os achados clínicos e radiográficos podem estar presentes desde o nascimento e o odontopediatra tem papel fundamental no diagnóstico precoce da síndrome de Ellis-van Creveld, bem como na prevenção de problemas bucais, reabilitação e intervenções estéticas.

Palavras-chave: cardiopatias congênitas, cárie dentária, odontopediatria, Síndrome de Ellis-van Creveld.

Introduction

Ellis-van Creveld (EVC) syndrome also known as chondroectodermal dysplasia is a rare autosomal recessive congenital disorder[1,2] caused by mutations in the two genes EVC1 and EVC2 localized in locus 16 on the short arm of chromosome [4.3] Short limb dwarfism, postaxial polydactyly of the hands, ectodermal dysplasia may affect hair, teeth and nails[1,4,5] and congenital heart malformations[1,4,5], present in about 50-60% of cases[6-8] are attributions that characterize the EVC syndrome.

It is estimated in the general population the prevalence of 7 in 1 million live births with no gender or race predilection.[6,9] Parental consanguinity may be present[2,9,10] being estimated in about 30% of the cases.[6]

EVC syndrome shows a wide spectrum of oral and dental manifestations, including: tooth agenesis, conical teeth[2,4,9], taurodontism [2,9-11], enamel hypoplasia[5,9,11], malocclusion [9,12,13], presence of multiples and accessories labial frenulum[2,9,11,13] and fusion of the upper lip anterior portion to the maxillary gingival mucosal margin.[2,9,11]

This article describes classic and rare oral manifestations of a Brazilian boy diagnosed with EVC syndrome. It is expected based on the findings of this case report that it can be assisted health professionals, especially pediatric dentists, in syndrome diagnosis, dental planning, and treatment decision-making, based on the best scientific evidence. Informed consent was obtained and signed by the child's parents.

Case Report

A 6-year-old boy attended in Oral Health Program at the Department of Pediatric Dentistry at the State University of Londrina, Brazil, complaining of aesthetic damage due to tooth agenesis. The child is the first and only child of non-consanguineous parents who reported no family history or characteristics of EVC syndrome. However, the parents reported that the child's maternal grandparents were first cousins.

1. Medical history:

a) Gestational and childbirth history: previous diagnosis of suspected syndrome on ultrasonography at the 34th gestational week, in which short bones characteristic of dwarfism and pulmonary hypoplasia were detected; cesarean delivery at 40 weeks, Apgar 7 and 8, weighing and measuring, respectively, 3.155 kg and 45 cm. According to the parents, the pregnancy and delivery were uneventful and there was no exposure to radiation during pregnancy.

b) Syndrome diagnosis: patient was diagnosed with EVC syndrome at 35 days of life. The diagnosis was made through the classical clinical characteristics of the patient: shortened limb dwarfism, postaxial polydactyly of the hands, ectodermal dysplasia and congenital heart malformations.

c) Neonatal period: intensive care unit monitoring for 55 days due to pulmonary hypertension, cardiac atrial septum defect, persistent left superior vena cava, bicuspid aortic valve, tricuspid insufficiency, and coarctation of the aorta. He performed heart surgery at 56 days of life, with good prognosis.

2. General characteristics: disproportionate short stature (112 cm), weight 21 kg, syndromic face, low-set ears, convergent strabismus and normal hair appearance (Figure 1), polydactyly affecting both hands, hypoplastic fingernails, thin, and spoon-shaped (Figure 2). No polydactyly was observed on the feet (Figure 3).

3. Previous dental history: at 3 months and 21 days of life, the patient underwent extraction of two maxillary neonatal teeth (51 and 61) due to uncommon ectopic implantation in soft tissue, conical-shaped crowns and hypoplastic enamel, in addition to difficulty during breastfeeding and consequently weight gain. Extraction was performed after antibiotic prophylaxis with amoxicillin oral suspension without intercurrences.

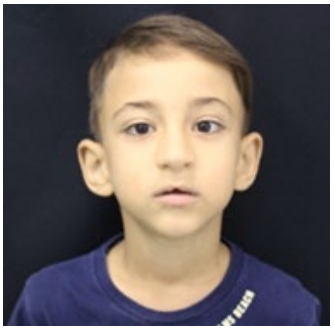


Figure 1. *Face syndrome showing the presence of low-set ears, convergent strabismus, and normal capillary appearance*

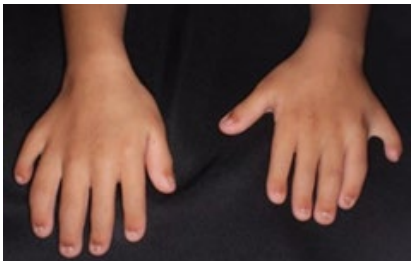


Figure 2. *Polydactyly, hypoplastic and spoon-shaped nails affecting both hands*

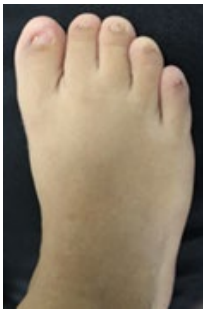


Figure 3. *Absence of polydactyly on the feet*

4. Oral characteristics: dental agenesis of teeth 52, 62, 71, 72, 81, 82, 11, 12, 21, 22, 31, 32, 33, 41, 42 and 43, taurodontism in all deciduous molars and certain degree of taurodontism of the maxillary and mandibular first permanent molars, delayed tooth eruption (Figure 4) and dental caries (Figures 5a and 5b). In addition, conical teeth, enamel hypoplasia, bilateral posterior crossbite, lower canine rotation, multiple accessories labial frenulum, absent vestibular sulcus, alveolar serrations in maxillary anterior region, and a whitish nodule located on the alveolar crest in the lower incisors region (Figure 6). Preventive oral hygiene measures, dental prophylaxis and application of cariostatic in the primary molars were procedures carried out in a first stage.

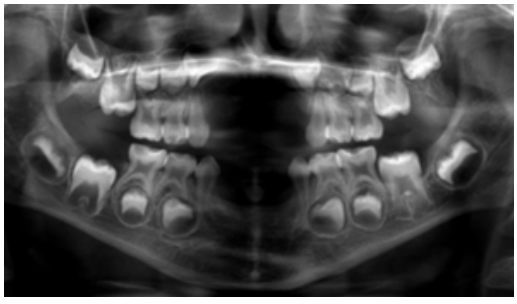
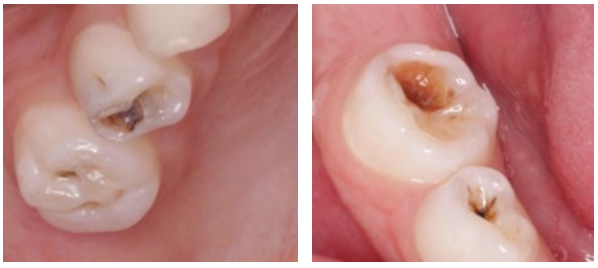


Figure 4. *Panoramic radiograph showing dental agenesis of the maxillary and mandibular incisors and permanent mandibular canines. Taurodontism affecting the deciduous maxillary and mandibular molars. A certain degree of taurodontism affecting the maxillary and mandibular permanent molars*



Figures 5a and 5b. *Presence of dental caries*

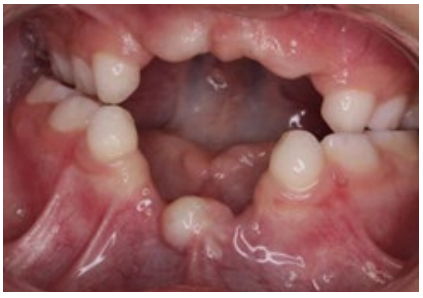


Figure 6. *Presence of multiple labial frenulum and absent vestibular sulcus, missing of teeth 52, 62, 71, 72, 81, 82, conical teeth, gyroversion of the lower canines, malocclusion and a nodule*

Discussion

EVC syndrome is a rare autosomal recessive congenital disorder[1,2] commonly found in the Amish community in the state of Pennsylvania, United States.[14,15] In the present report, the patient has a history of consanguineous marriage in the family, a fact that supports the syndrome recessive character. However, the patient is of Brazilian origin with no family history of EVC syndrome.

The patient had the EVC syndrome classic tetrad: disproportionate short stature, polydactyly in both hands, alterations affecting teeth shape/size, dysplastic nails and congenital heart malformation. According to Digilio et al., heart defects represent the main causes of reduced life expectancy.[7] Therefore, it is necessary antibiotic prophylaxis to prevent bacterial endocarditis16 before preventive dental procedures. Furthermore, the present report is the first clinical case of EVC syndrome associated with pulmonary hypoplasia.

Classic oral and dental characteristics found in this patient corroborate those

described by several authors: teeth agenesis, conical teeth[2,9,12], enamel hypoplasia[5,9,11], natal/neonatal teeth[9,10], malocclusion[9,12,13], presence of multiples accessories labial frenulum and fusion of the upper lip anterior portion to the maxillary gingival mucosal margin.[2,9,11] According to Lauritano *et al*.[17], among the manifestations mentioned, the presence of conical teeth is the most prevalent (61.4%), followed by dental agenesis (47.7%), multiples accessories labial frenulum (45.5%), enamel hypoplasia (38.6%), fusion of the upper lip anterior portion to the maxillary gingival mucosal margin (13.6%), natal/neonatal teeth (9.1%), and crossbite (6.8%).[17] However, in the literature, there are reports of unusual findings, such as: taurodontism[2,9,11], supernumerary tooth[11,18], microdontia, dens in dente, talons cusp[2,4], single-rooted deciduous molars[19], impacted tooth[2] and dental transposition.[5] In this clinical case, the presence of taurodontism was detected in the deciduous molars and a certain degree of taurodontism in the permanent molars. In addition, the patient had a high prevalence of dental caries, a disease that can be explained by alterations in dental anatomy and hypoplasia.[2,5] Nonetheless, parents reported a diet with a high frequency of sugar intake and difficulty in controlling dental biofilm effectively. Table I presents and compares the main manifestations of EVC syndrome.

Another aspect reported by Aminabadi, Ebrahimi, Oskouei[11] and Hunter and Roberts[6] refers to talkativeness. This child's communicative profile was clearly observed during dental care and may be a behavioral characteristic of patients diagnosed with EVC syndrome. Thus, the child's capacity for

Table 1. *Oral manifestations of EVC syndrome in the literature compared to the clinical case.*

ORAL MANIFESTATIONS REPORTED IN THE LITERATURE	ORAL MANIFESTATIONS IN THE PATIENT
Natal/neonatal tooth	Present
Supernumerary tooth	Not present
Teeth agenesis	Present
Conical teeth	Present
Dens in dente	Not present
Microdontia	Not present
Talon cusps	Not present
Single-rooted molars	Not present
Taurodontism	Present in deciduous molars and presence to a certain degree in permanent molars
Enamel hypoplasia	Present
Dental caries	Present
Impacted tooth	Not present
Dental transposition	Not present
Rotated tooth	Present
Bilateral posterior crossbite	Present
Delayed tooth eruption	Present
Serrations of the alveolar ridge	Present
Vestibular sulcus	Present
Multiple labiogingival frenula	Present
Midline notch in upper lip	Present
Hypertrophy of labiogingival frenula	Not present
High arched palate	Not present
Macroglossia	Not present

social, dialogic and expressive interaction represented an important factor in the establishment of bonds and effective dental behavioral management.

Measures for the prevention and control of dental caries, such as dietary counseling, dental biofilm evidence, oral hygiene instruction, prophylaxis and dental restorations represent relevant strategies to promote health that have been implemented. At a later stage, these indispensable procedures for the maintenance of oral health, aesthetic and functional rehabilitation, and orthodontic intervention will be used to correct malocclusion, speech, chewing, and improve quality of life.

Conclusion

EVC syndrome represents a rare autosomal recessive disorder with varied oral and general manifestations that require a multidisciplinary approach. Pediatric dentists have an essential role in the early diagnosis of oral characteristics that can be present from birth. Besides, an effective treatment plan in terms of oral health prevention, satisfactory rehabilitation and aesthetics interventions must be carried out to improve the patient's quality of life.

Conflict of Interest

The authors declare that there are no conflicts of interest regarding the publication of this paper.

Ethics Statement

The authors declare that the parents gave consent for images and clinical information of the case to be reported in scientific publications. Parents understand that the child's name and initials will not be published and efforts will be made to conceal the child's identity. This article complies with the protocols of the Research Ethics Committee of the State of University of Londrina.

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