Ellis-van Creveld syndrome: systemic and oral findings

Síndrome de Ellis-van Creveld: achados sistêmicos e orais

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ABSTRACT

Aims: To present a case of Ellis-van Creveld syndrome highlighting the systemic and oral manifestations and expand the phenotypic spectrum of the disorder.

Case description: A 4-year-old female, with an initial diagnosis of Ellis-van Creveld syndrome, was referred for dental treatment. The patient had hexadactyly of the hands, thorax disproportionate to the shortened limbs, hypopigmented and dry skin, short stature, a wide gap between the 1st and 2nd toes of the right foot and markedly dystrophic toenails. The oral manifestations were upper labial frenulum causing large vestibule and absence of diastema, labiogingival frenulum, enamel hypoplasia, conical teeth, claw-like cusp, oligodontia, microdontia and malocclusion.

Conclusions: Ellis-van Creveld syndrome is one among several syndromes with oral manifestations that demands the participation of a multidisciplinary team to better patient assessing, treatment and monitoring. Dentists have the leading role in the control and treatment of the oral manifestations.

KEY WORDS: ELLIS-VAN CREVELD SYNDROME; CHONDROECTODERMAL DYSPLASIA; HYPODONTIA; ANODONTIA; CASE REPORT.

RESUMO

Objetivos: Apresentar um relato de caso da síndrome de Ellis-van Creveld, destacando as manifestações sistêmicas e orais e expandindo o espectro fenotípico da doença.

Descrição do caso: Uma menina de 4 anos com diagnóstico inicial de síndrome de Ellis-van Creveld foi encaminhada para tratamento odontológico. A paciente apresentava hexadactilia das mãos, tórax encurtado e desproporcional aos membros, cabelo fino, pele hipopigmentada e seca, baixa estatura, espaçamento entre o primeiro e o segundo dedos do pé direito e unhas dos pés acentuadamente distróficas. As manifestações orais foram representadas por freio labial superior amplo causando ausência de vestíbulo e diastema, frênulos labiogengivais, hipoplasia do esmalte, dentes cônicos, cúspide em garra, oligodontia, microdontia e maloclusão.

Conclusões: A síndrome de Ellis-van Creveld representa uma entre as diversas síndromes com manifestações orais que demandam a participação de uma equipe multidisciplinar para melhor avaliar, tratar e acompanhar os pacientes. O cirurgião dentista tem o principal papel no controle e tratamento das manifestações orais.

DESCRITORES: SÍNDROME DE ELLIS-VAN CREVELD; DISPLASIA CONDROECTODÉRMICA; HIPODONTIA; ANODONTIA; RELATO DE CASO.

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INTRODUCTION

Ellis-van Creveld syndrome (EvC), OMIM 225500, is a rare autosomal recessive skeletal dysplasia with a birth prevalence of 7/1,000,000,000 for approximately 300 cases reported so far.[1] It has inter and intrafamilial variability and is characterized by chondrodysplasia, postaxial polydactyly, ectodermal dysplasia, and congenital malformations.[1-4] In about two thirds of patients, mutations in either EVC or EVC2 genes, located on chromosome 4p16, have been found to be the underlying cause of EvC.[5-9]

Oral manifestations include submucous clefts, absent vestibular sulcus, multiple musculofibrous frenula, dystrophic philtrum, hypodontia, enamel hypoplasia, delayed eruption, natal teeth, microdontia, conical teeth, dens in dente, taurodontism, high caries rate and malocclusion, besides diastema.[1-3,10-12]

Herein we report a case of EvC with systemic and oral manifestations, highlighting the importance of a multidisciplinary therapeutic planning and the fundamental role of the dentist in its early diagnosis and treatment.

CASE REPORT

This case report was approved by the Ethical Research Committee of the University of Alfenas (UNIFENAS) (79/2008).

A 4-year-old female with the diagnosis at birth of Ellis-van Creveld syndrome was referred for regular dental treatment. There was no consanguinity and pregnancy was uncomplicated. She was the fourth daughter of clinically normal parents and siblings. Other family members did not present a similar condition. The patient was born with hexadactyly of the hands, a narrow thorax and disproportionate small stature with increasing severity from the proximal to distal portions of the limbs (Fig. 1A, B). No cardiac abnormalities were found. Other abnormalities were diagnosed in her first few years of life, including thin hair, hypopigmented and dry skin, retarded growth, a wide gap between the 1st and 2nd toes of the right foot and markedly dystrophic fingernails and toenails (Fig. 1C). She also presented asthma, bronchitis, and difficulty in walking due to the abnormal shape of the legs. At the time of our examination, abdominal and renal ultrasound, echocardiogram and electrocardiogram were normal. Moreover, all biochemical and hormone blood and urine tests were normal. She presented normal intelligence quotient.

Extra-oral examination revealed hypertelorism and a mild defect in the labial philtrum. Intra-oral examination showed a very shallow upper labial sulci due to the presence of a broad labial fraenum largely obliterating the labial sulcus, multiple labiogingival frenula, enamel hypoplasia, conical teeth, and diastema (Fig. 2A). Radiographic examination showed dental abnormalities such as oligodontia, microdontia, moderate taurodontism, bicuspid teeth, and irregular spacing (Fig. 4B, C). The patient presented good oral hygiene. Skeletal radiography revealed osseous syndactyly of the 5th and 6th metacarpals of the left hand, clinodactyly of the 6th finger, hypoplastic phalanges, metacarpals and metatarsals of the hands and feet, and disturbance in bone modeling of the tibia (distal extremity). Genu valgum was present, as a result of the abnormal tibial epiphysis. (Fig. 1A). The patient had also hypoplastic radius and ulna, humerus valgum and mild scoliosis (Fig. 3A).

Initial dental treatment consisted of oral hygiene orientation. Fissure sealants and restorations were performed. Orthodontic, prosthetic and surgical therapies are currently under consideration.

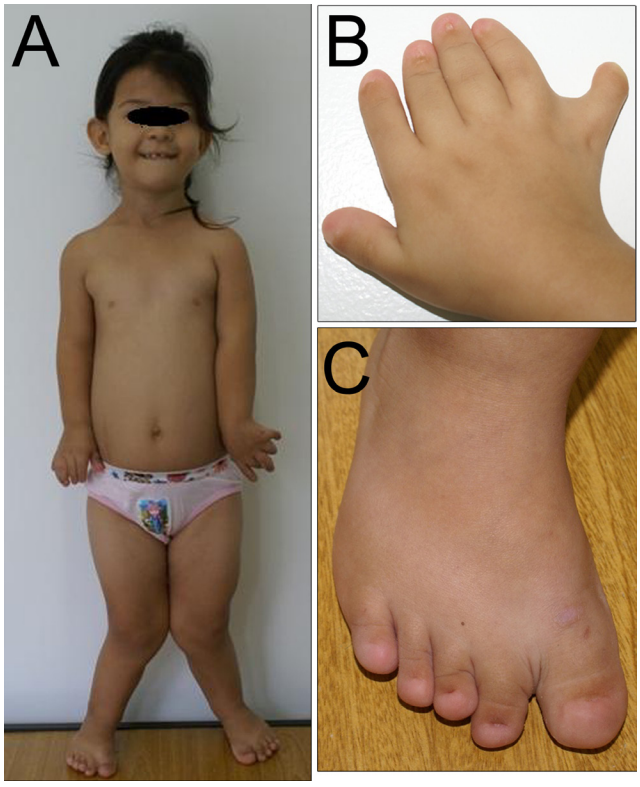

Figure 1. (A) Frontal view of the patient with a narrow thorax and disproportionate small stature and genu valgum. **(B)** Hexadactyly of the hands and dystrophic nails. **(C)** Markedly dystrophic fingernails and toenails and a wide gap between the 1st and 2nd toes of the right foot.

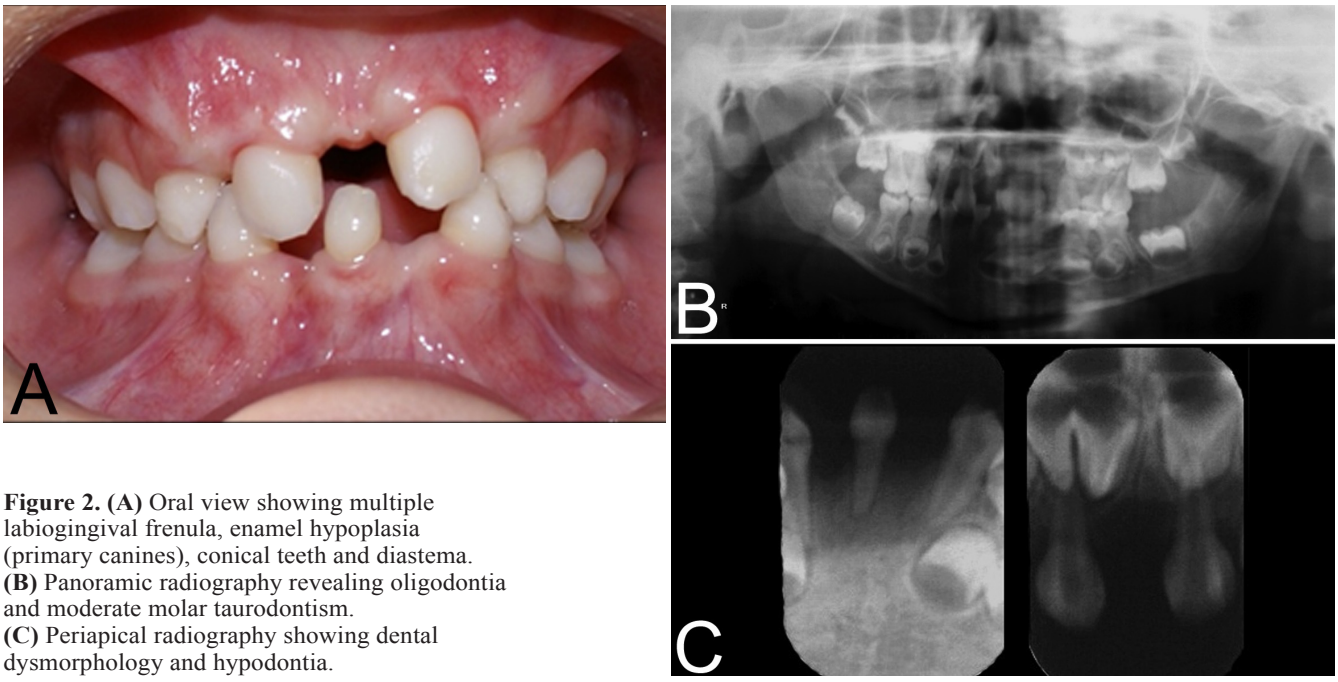

Figure 2. (A) Oral view showing multiple labiogingival frenula, enamel hypoplasia (primary canines), conical teeth and diastema. **(B)** Panoramic radiography revealing oligodontia and moderate molar taurodontism. **(C)** Periapical radiography showing dental dysmorphology and hypodontia.

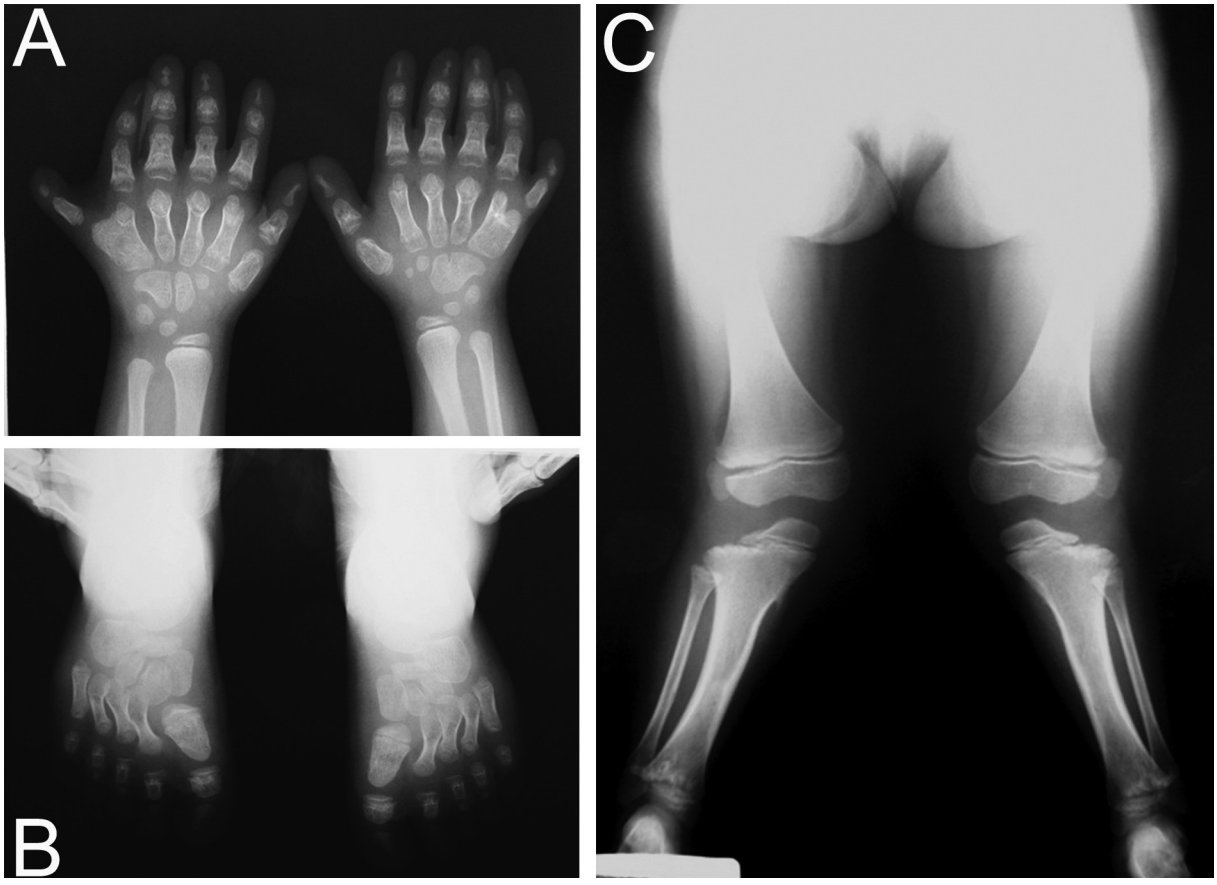

Figure 3. (A, B, C) Skeletal radiography revealed osseous syndactyly of the 5th and 6th metacarpals of the left hand, clinodactyly of the 6th finger, hypoplastic phalanges, metacarpals and metatarsals of the hands and feet, and disturbance in bone modeling of the tibial zone (distal extremity).

DISCUSSION

Our patient showed several clinical features that characterize EvC syndrome: bilateral polydactyly affecting hand, ectodermal dysplasia affecting skin, hair, nails and teeth, genu valgum and disproportionate small stature. The extremities were plump and progressively shortened in the distal regions. A narrow thorax with short poorly developed ribs gave rise to a pigeon breast appearance.

Although congenital heart malformations occur in 50-60% of cases,[1,3,4] they were not found in the present case. Several additional clinical findings that have been reported, including strabismus, hypospadias, cryptorchidism, thoracic and pulmonary malformations and renal abnormalities,[1,3,4] were not present in this case.

Fusion of the middle portion of the upper lip to the maxillary gingival margin that eliminates the maxillary labial vestibule, and the presence of numerous frenula tethering the upper lip to the gingiva are consistent features of EvC, [2,3,10-12] and were identified in our case.

EvC patients may show numerous dental abnormalities.[1-3,10-12] Our patient has several dental abnormalities in terms of number (oligodontia), size (microdontia of the canines and incisors) and shape (conical molars, canines and incisors and bicuspid incisors). Moderate taurodontism is also present in molars deciduous.

EvC syndrome requires a multidisciplinary therapeutic approach i.e., orthopedic corrections, surgical repair of cardiac malformations, and dental intervention for several dental abnormalities.[3,9,13,14] Dental treatment includes oral hygiene instructions and preventive professional care.[12] Concerning the malformed teeth, construction of prosthetic crowns and partial or fixed denture (considering age) are indicated.[12,14,15] Hypoplastic teeth and decayed teeth can be restored with composite restorations for better esthetics and preservation of tooth structure.[14,16] These patients may need orthodontic treatment[12] and dental implants can be studied in each particular case when a patient reaches adulthood.[1,15] In the present case the regime included oral hygiene orientation, the use of fissure sealants, and restorations. The maintenance of a caries-free permanent dentition as well as the provision of orthodontic, prosthetic and surgical therapy are necessary and will be provided.

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