Ellis-van Creveld syndrome: oral manifestations and treatment

Síndrome Ellis-van Creveld: manifestações bucais e tratamento

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Abstract

The Ellis-van Creveld syndrome (EVC) or chondroectodermal dysplasia is a rare autosomal recessive disease, with incidence of 1:244,000 for the general population. The exact prevalence is unknown, but believed to be a genetic chromosomal disorder and a higher incidence in women. EVC consist of a tetrad of principal features: chondroectodermal dysplasia, polydactyly, congenital heart defects, and hypoplastic nails and teeth. In this syndrome many other organs can be compromised and other oral manifestations may be present. We report a case of a patient, 17 years old, female, presenting the typical features of EVC and the medical report confirming the condition. Attended at the Center for the Studies and Care of Special Patients of University Paulista – São Paulo, Brazil, seeking dental treatment, reported being dissatisfied with the aesthetic. We analyze the events and the patient's oral condition and its treatment recommended. This study was previously submitted and approved by the University Paulista Ethic Committee with the protocol nº 642/09.

Descriptors: Ellis-van Creveld syndrome; Polydactyly; Dwarfism; Heart septal defects; Dental enamel hypoplasia

Resumo

A síndrome de Ellis-van Creveld (EVC) ou displasia condroectodérmica é uma rara doença autossômica recessiva , cuja incidência na população em geral é de 1:224.000. A prevalência exata é desconhecida, acredita-se ser uma desordem genética cromossômica e de maior incidência em mulheres. A EVC é dada pela presença tétrade característica: displasia condroectodérmica, polidactilia, defeitos cardíacos congênitos e unhas e dentes hipoplásicos. Além da tétrade, muitos outros órgãos podem ser comprometidos e outras manifestações bucais podem se fazer presentes. Relata-se um caso clínico de uma paciente, 17 anos, genêro feminino, apresentando as típicas características da EVC e confirmando com laudo médico a patologia. Compareceu ao Centro de Estudos e Atendimento a Pacientes Especiais da UNIP em busca de tratamento odontológico e relatou estar insatisfeita com a estética. Foram analisadas as manifestações e condições bucais da paciente e seu tratamento preconizado.

Descritores: Síndrome de Ellis-van Creveld; Polidactilia; Nanismo; Defeitos dos septos cardíacos; Hipoplasia do esmalte dentário

Introdução

Syndrome Ellis-van Creveld (EVC) or dysplasia chondroectodermal was described in 1940 by Richard Ellis and Simon van Creveld, as an autosomal recessive disorder due to a genetic defect located on chromosome 4p16[1].

The incidence of EVC syndrome is 1:244,000 of the population, around 150 cases are described in the literature and is more common in closed ethnic communities as the Amish population of Pennsylvania, with an incidence of 2:1,000[2]. Almost half of these patients die during childhood by cardiopulmonary defects. For that reason the life expectancy of patients with EVC is determined by their congenital heart disease[3-4].

The diagnosis can be made during the prenatal period from the 18th week of gestation by ultrasonography and later by clinical examination after birth, when is noted the presence of tetrad features: chondroectodermal dysplasia, polydactyly, cardiac defects and congenital hypoplastic teeth, and the pseudo cleft lip and no groove-gingival sulcus[2,5]. It is believed that it results from a genetic defect located on chromosome 4p16[6]. However, the clinical presentation is variable and the whole spectrum can be devoid of any patient[3,7].

Ectodermal dysplasia affects the bones and causes a severe lack of ossification, thus generating the shortening of the ends of limbs and stature, with an option of treatment of distraction osteogenesis to lengthen the members and in relation to treatment with growth hormone, there is no scientific evidence of improvement in the literature[7-9]. The involvement of polydactyly occurs in hands and feet, changing the shape and number. The fingers are in a "sausage" form and nails are hypoplastic[2,10].

The chest is often narrow with pectus excavatum, lumbar lordosis and knee valgum, the hair is sparse and thin[8].

Congenital heart defects are described in 50 to 60% of patients affected by this syndrome. The main defects occur in the mitral and tricuspid valve and the atrial and ventricular septal responsible for the decrease in life expectancy in these patients[1,5,7,11]. For the cardiac defects observed in patients with EVC is necessary to evaluate the antibiotic coverage for the prevention of infective endocarditis, where the procedures to be performed at the dental office are surgical trauma[10,12].

The oral manifestations are diverse and involve not only the soft tissues, but also the number, shape, and structure of the teeth. The most common finding is a fusion of the anterior portion of the upper jaw to the gingival margin, eliminating the groove between the mucosa and maxillary marginal gingiva[3,11,13-14]. The anterior portion of the lower alveolar ridge is often serrated, and several small lip brakes are frequent. The teeth tend to be small and conical; molars may have abnormal cusps or additional slots and sometimes hypoplastic enamel. Congenital oligodontia of the deciduous and permanent teeth, presence of supernumerary teeth, natal and neonatal roots dysmorphic and delayed eruption were also reported[10-11,13].

The prevention of holes development is necessary, as a guideline for diet, plaque control, oral hygiene instructions and applying fluoride varnish or mouthwash daily. Decayed teeth may receive the restoration technique with classical glass ionomer and amalgam or composite resin or an atraumatic restoration technique (ART). Concerning the micro dents or peg-shaped teeth, is effective the construction of prosthetic crowns and concerning the anodontia, partial removable prosthesis or bridge may be used to recover occlusion, mastication and esthetics. These patients may need orthodontic treatment, malocclusion may occur without skeletal pattern, but in the course of the abnormalities[3,10,14-15]. The use of dental implants can be studied in each particular case when a patient reaches adulthood and has a stabilization of the bone area[9]. The dental ma-

nagement depends on each case and requires a multidisciplinary team of medical geneticists, speech therapists, physical therapists, surgeons, dermatologists and specialized dentists.

This study aims to report the case of a patient with Ellis-van Creveld, its general features, the oral findings and treatment are contributory factors to dentists, to improve its diagnosis.

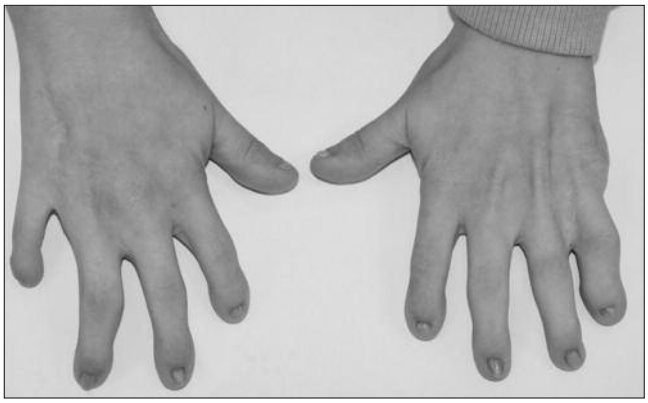

Figure 1. Hands of the patient after surgical correction with small changes, where the fingers are short and curved

Figure 2. Photo front of the patient, where it is noticed the short stature, curvature of the arms and posture

Case report

The patient A.C.P.A., 17 years old, female, attended at the Center for the Studies and Care of Special Patients at UNIP, seeking dental treatment, reported being dissatisfied with the aesthetic. She reported being the daughter of a consanguineous marriage and was reported in her medical history, the EVC syndrome. In childhood the polydactyly of the hands and feet "Y" have been corrected surgically, but the physical examination pointed a remarkable the change that affects the motor coordination and the oral hygiene (Figure 1).

There is no change in facial morphology (Figure 2), however, is present short stature, the hair is fine and don't have quantitative change, the nails are hypoplastics and presents hypotrichosis of the eyebrows. Also in the anamnesis was reported that prenatal and neonatal teeth were observed, with delayed eruption of other dental elements.

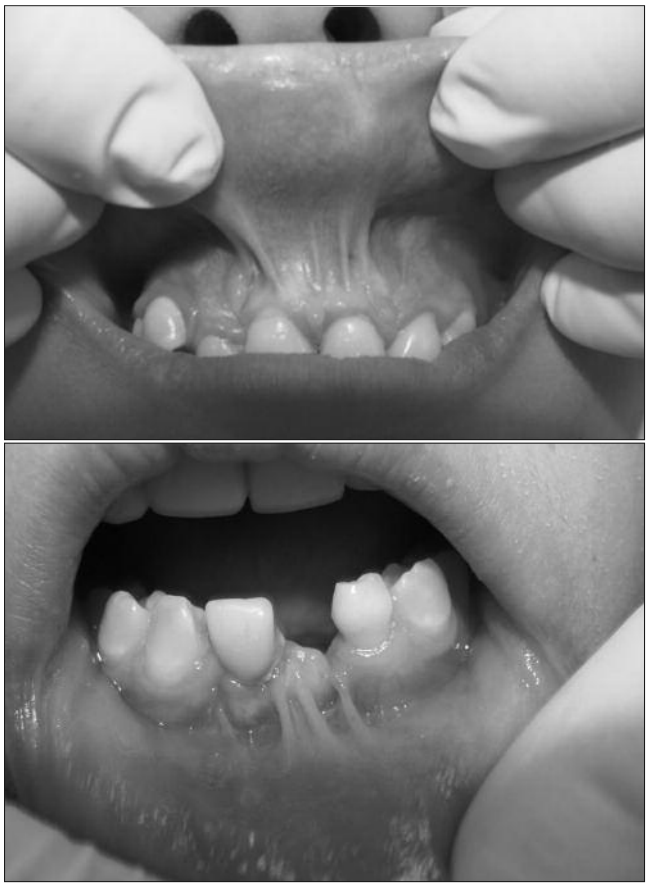

Figure 3. Multiple lower lip brakes and upper lip extended brak

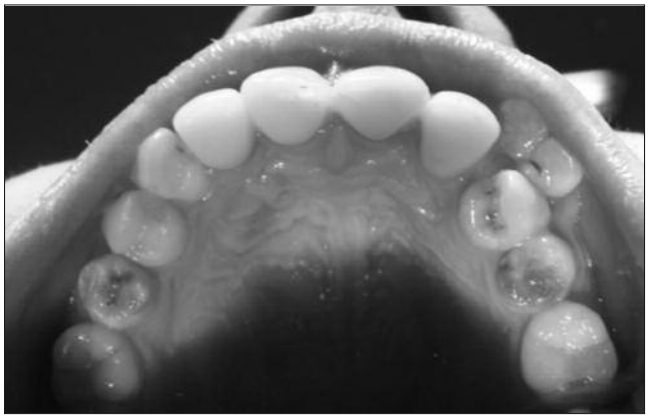

Figure 4. Photo of occlusal upper of the patient, where it is noticed the presence of supernumerary fused, hypoplastic spots in the enamel

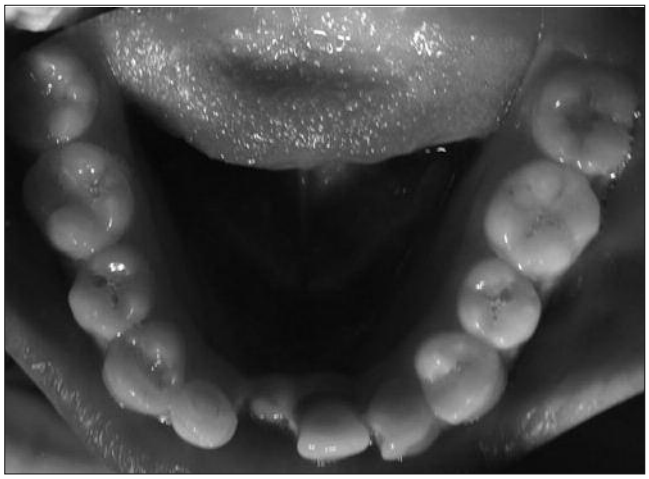

Figure 5. Photo of occlusal lower patient, where it is observed agenesis of the lower central incisor

As for the oral soft tissues, there are multiple lower lip brakes and upper lip extended brake (Figure 3). In the mouth there is agenesis of the elements 41, 43, 31 and 37, presence of supernumerary elements between 12 and 13, peg-shaped teeth and enamel hypoplasia (13,14,25,33,34 and 47) (Figures 4 and 5). It was observed high rate of plaque (66%) and presence of active carious lesions (36 and 46). Salivary test was performed (Dentobuff® - Inodon), where it was found: oral pH remained neutral[4,6], buffer capacity high and normal salivary flow.

The treatment is based on adapting the oral environment, removing the caries and restoring them. Was indicated and performed the surgical removal of supernumerary, enabling a future orthodontic treatment. Also, it was decided to rehabilitate elements engaged in aesthetics and function. The patient received preventive measures to guide diet, plaque control, oral hygiene instructions and fluoride varnish application.

Discussion

According to Krakow *et al.*[14] (2000) and Ruiz-Perez and Goodship[4] (2009), it is believed that the EVC is the result of a genetic defect located on chromosome 4p16, and being common the presence of endogamy and consanguinity.

Hunter and Roberts[10] (1998) state that about 30% of EVC cases the individuals are breeds from consanguineous relations. In this study, the patient says to be daughter of a consanguineous marriage of parents, cousins, and the continuous discovery of new chromosomal abnormalities confirms once more the genetic heterogeneity of the syndrome[3].

No need to study and research of genetic defect on chromosome 4p16 for diagnosis of the syndrome[6-7,12], because it is essentially clinical. There is a need for additional tests such as radiology, laboratory and cardiac function, in that the changes are found in physical examination of the patient. Ectodermal dysplasia affects the tubular bones producing a serious defect of ossification, thus generating the shortening of the ends of limbs and short stature[7,9,15]. Baujat *et al.*[5] (2007) reported that the bone growth is delayed, which culminates in the short stature of these individuals, which was also observed in this report.

According to Pinto Jr. *et al.*[15] (2003), Hunter and Roberts[10] (1998) and Kurian *et al.*[13] (2007), the polydactyly is universal in the syndrome of Ellis-van Creveld and usually is post-axial, occurring most often the ulnar side of the hands. The feet are less affected. The syndactyly is seen in some cases and may occur with carpal mergers and changes in the shape of the phalanges. The case report presented polidactyly and syndactyly, but both were corrected in childhood, interfering in the fine motor coordination.

Cahuana and Palma[3] (2004) reported that approximately 50% of these patients die during childhood by cardiopulmonary complications; therefore, the life expectancy of patients with EVC is determined by their congenital heart disease. In the case presented, the patient underwent surgical correction of heart in childhood and show no complication or defect.

Conclusion

We conclude that the dental surgeon has an important role in early diagnosis and establishing treatment protocols (aesthetic and functional) that improves the quality of life of patients and establishing a differential diagnosis with other pathologies.

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