CASE HISTORY REPORT

ABSTRACT

Ellis-Van Creveld is a rare syndrome with characteristic dental and orofacial findings. Dental management of patients with Ellis-Van Creveld syndrome can be complicated by the associated skeletal and cardiac abnormalities. Here, we present the dental and orofacial findings in a patient with Ellis-Van Creveld syndrome, describe a new oral finding, and discuss the dental management considerations.

KEY WORDS: Ellis Van Creveld, chondroectodermal dysplasia, rare diseases

Ellis-Van Creveld syndrome: dental management considerations and description of a new oral finding

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Spec Care Dentist 35(6): 312-315, 2015

Introduction

Ellis-Van Creveld (EVC) syndrome, also termed chondro-ectodermal dysplasia, is a rare autosomal recessive disorder first described in 1940 by Richard W. B. Ellis of Edinburgh and Simon van Creveld of Amsterdam.[1] Medical historians have suggested that King Richard III had features of EVC syndrome based on Sir William Shakespeare's and Sir Thomas More's descriptions of his deformities, but recent archaeological discoveries in Leicester have precluded this.[2–4]

The prevalence of EVC is not exactly known, but is estimated to be 7 per 10^6 population.[5] The syndrome is most prevalent (1/5,000 live births) in the Old Order Amish communities of eastern Pennsylvania, United States, as a consequence of continuing intermarriages.[6]

The phenotypic features of EVC syndrome, due to mutations of the *EVC1* and *EVC2* genes located on chromosome 4p16 include short stature, polydactyly, dysplastic hair and nails, congenital heart defects, and oro-dental abnormalities.[7,8] The latter include natal teeth, hypodontia, enamel hypoplasia, malocclusion, malformed teeth, fusion of lips and labial gingivae, and hypertrophy of labiogingival fraenum.[9]

Recognition of oral features of EVC syndrome is important to differentiate it from similar chondrodystrophies such as Jeune dystrophy, McKusick-Kaufman syndrome, achondroplasia, and Weyers acrodental dysostosis.[5,10,11]

Special care dentists might be consulted regarding oral manifestation and dental treatment planning of patients with EVC syndrome. We present a new case of EVC syndrome, describe its oral and dental findings, and discuss the dental management considerations for patients affected by this rare syndrome.

DOI: 10.1111/scd.12116

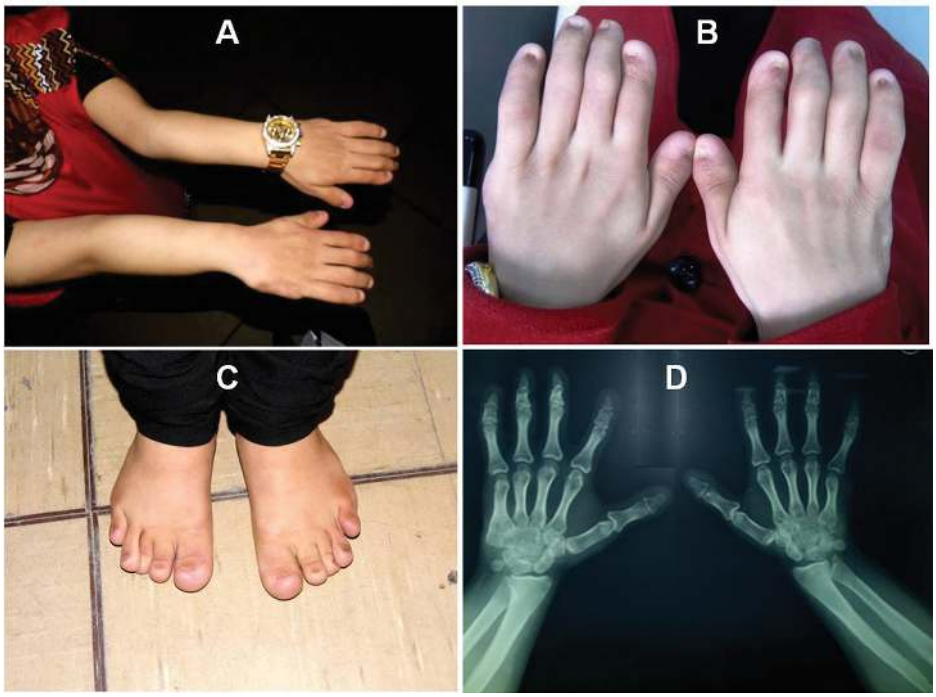

Figure 1. Limb deformities observed in our patient. (A) Outward bending of elbows (cubitus valgus), (B) short and sausage-shaped fingers with hypoplastic nails; (C) small feet, syndactyly and hypoplastic nails; (D) X-ray of both hands.

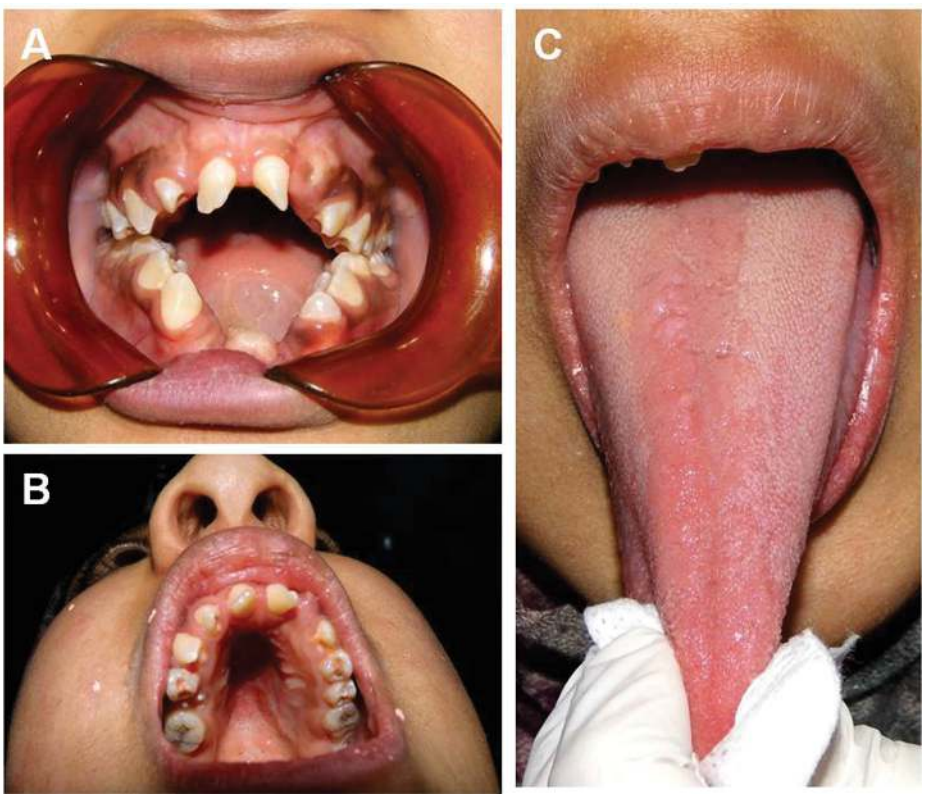

Figure 2. Oro-dental abnormalities include (A) Hypodontia, spacing, conical-shaped upper incisors, anterior open bite, and enamel hypoplasia particularly on molars and; (B) high arched palate; (C) long and pointed tongue.

Case report

A 17-year-old girl presented for orthodontic treatment. The patient was the third child of consanguineous (paternal cousins) parents. Her birth was at full term, a normal delivery with birth weight around 2.7 kg. Her mother reported that the girl was borne with a tooth in the lower labial segment (natal tooth). Her mental development was within normal limits and she had recently completed her high school education.

Upon general examination, the patient had disproportionately short stature (139 cm) and weighed 34 kg. Examination of her hands showed short sausage-shaped digits, hypoplastic nails, and scars next to both the little fingers as a result of previous surgical removal of an extra digit from each hand (Figure 1). Both arms were deformed with outward bending of elbows (cubitus valgus; Figure 1). She also had small feet, syndactyly, and hypoplastic nails (Figure 1). Hair, sebaceous and sweat glands were normal, and chest radiography was normal.

Extraoral examination of the head and neck showed mild strabismus and mandibular protrusion and deviation to the left side. Intraoral examination revealed an anterior open bite, cross bite in the left buccal segment, high arched palate, and rudimentary development of the lower labial segment (Figure 2). Dental examination showed hypodontia of all lower incisors, impacted upper left lateral incisor and a partially erupted upper second premolar, conical shaped upper central incisors, and enamel hypoplasia particularly evident on molars and premolars (Figure 2). OPG examination revealed taurodontism anomaly of molar teeth and impaction of lower third molars (Figure 3). Soft tissue examination revealed leukoedema on both buccal mucosae, ethnic pigmentation on upper and lower gingivae, and fibrous hypertrophy of the lower labia frenum (Figure 2). Interestingly, the tongue appeared long and pointed with linear streaks devoid of papillae (Figure 2).

A multidisciplinary treatment approach involving restorative dentistry, maxillofacial surgery, and orthodontics

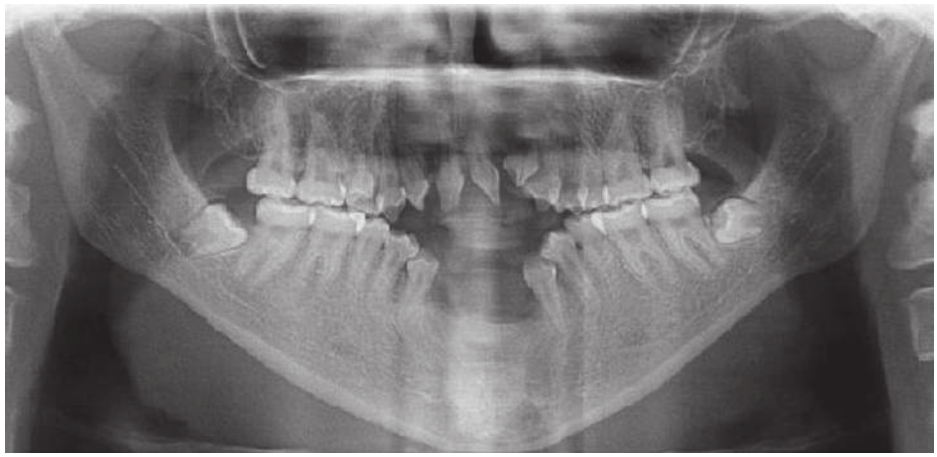

Figure 3. Panoramic radiograph showing hypoplasia of all lower incisors, impaction of upper left lateral incisor, and taurodontism affecting upper and lower molars. The lower third molars appeared impacted.

was planned. Phase one of our treatment plan involved improving patient's oral hygiene practices, dietary counseling, and treatment of active dental diseases including occlusal amalgam fillings for teeth #14 & 30. Surgical exposure of the impacted upper lateral incisor was performed under local anesthesia. Surgical orthodontics was planned to correct the associated skeletal defects, mainly anterior open bite and mandibular protrusion and deviation. Orthodontic treatment involved leveling and alignment, dentoalveolar decompensation, and arch coordination. In addition, space analysis was performed to plan future prosthetic replacement of congenitally missing teeth using dental implants. Bimaxillary osteotomy, involving impaction of the maxilla and mandibular setback, is planned to be performed once the orthodontic phase is complete. In addition, genetic counseling was advised since the girl is approaching marital age and with consanguineous marriages particularly common in the Middle East.

Discussion

EVC syndrome is a rare autosomal recessive disorder caused by mutations of *EVC1* and *EVC2* genes located on chromosome 4p16.[7] Our patient demonstrated classical phenotypic features of EVC syndrome, namely the disproportionate short stature, limb deformities (cubitus valgus), polydactyly, dystrophic and hypoplastic nails, and characteristic oral and dental features. Congenital heart defects, such as atrial septal defect, ventricular septal defects, mitral and tricuspid valves defects, patent ductus arteriosus, and hypoplastic left heart syndrome, are reported in 50% to 60% of patients and are the main cause of death.[12] No signs of cardiac involvement were evident in our patient.

Rarely reported features in EVC syndrome include strabismus, epi- and hypospadius, cryptorchidism, thoracic wall and pulmonary malformation, thymic hypoplasia, and renal abnormalities.[5,13] Our patient had mild strabismus with no signs of renal, thoracic, or pulmonary abnormalities and demonstrated normal mental, psychological, and motor development—as do most patients with EVC syndrome.[5]

Our patient demonstrated characteristic orofacial features including, hypodontia, enamel hypoplasia, malformed teeth, neonatal teeth, taurodontism, malocclusion, fusion of lips and labial gingivae, multiple hypertrophic frenai, high arched palate, failure of eruption, and premature tooth loss.[14–20] Interestingly, her tongue was long and pointed, a feature not previously described in EVC syndrome. Further clinical and genetic studies however are needed to confirm any association between tongue abnormalities and EVC syndrome.

Recognition of oral and dental features in EVC syndrome provides important diagnostic information to distinguish this syndrome from other syndromes demonstrating similar phenotypic features such as Jeune dystrophy, McKusick-Kaufman syndrome, achondroplasia, and Weyers acrodental dysostosis.[5,11,21] Dentists therefore can have an important role in the diagnosis.

Diagnosis of EVC syndrome is based on family history, characteristic clinical, radiographic, and dental features, and molecular mutational sequencing. Prenatal diagnosis of EVC can be made using ultrasonography, which shows characteristic structural fetal defects in the late first trimester.[5]

Dental management of adult patients with EVC syndrome is complex and often requires a multidisciplinary approach to correct the associated dental defects. Maxillofacial surgery and orthodontics are often needed to correct malocclusion and manage unerupted teeth. Comprehensive restorative treatment is essential to manage enamel hypoplasia and to replace missing teeth. Our patient demonstrated severe malocclusion with a skeletal component, therefore a surgical orthodontic treatment was planned to correct mandibular asymmetry, anterior open bite, and unilateral cross bite. Dental caries and enamel hypoplasia were managed by restorations, fluoridated toothpaste, and dietary advice. The patient will be re-evaluated following completion of the orthognathic treatment regarding replacement of missing teeth and correction of dental aesthetics.

As a result of upper limb deformity and the inability to make a proper fist, some patients have difficulties in using toothbrush, and an adjustment of the toothbrush handle or the use of an electronic toothbrush might be appropriate.

Patients with congenital heart defects associated with EVC syndrome might need prophylactic antibiotics to prevent infective endocarditis before invasive dental procedures. The current U.K. National Institute of Clinical Excellence

(NICE) guidelines do not recommend prophylactic antibiotic to prevent infective endocarditis in patients with congenital heart defects.[22] However, guidelines from other bodies such as the American Heart Association (AHA) and the European Society of Cardiology (ESD) still recommend using prophylactic antibiotics to prevent infective endocarditis in some patients with congenital heart defects.[23,24] Dentists therefore are encouraged to consult the patient's cardiologist and to follow the guidelines applied in their region.[25]

Summary

EVC syndrome is a rare autosomal recessive condition with characteristic oral features. Recognition of oral and dental features provides important diagnostic information to distinguish this syndrome from other syndromes with similar phenotypic features. Dental management of patients with EVC syndrome requires a multidisciplinary approach to correct the associated dental and skeletal defects. Special care dentists might be consulted regarding oral manifestations and dental treatment planning for patients with EVC.

Conflict of interest

None declared.

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