Case Report

Chondroectodermal Dysplasia: A Rare Syndrome

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Abstract

Chondroectodermal dysplasia (Ellis-Van Creveld syndrome) is a rare autosomal recessive congenital abnormality. This syndrome is characterized by a spectrum of clinical findings, among which chondrodystrophy, polydactyly, ectodermal dysplasia, and congenital cardiac anomalies are the most common. It is imperative to not overlook the cardiac complications in patients with this syndrome during dental procedures. The case presented here, although quite rare, was detected under normal conditions and can be alarming for dental care providers.

Clinical reports outline the classical and unusual oral and dental manifestations, which help health care providers diagnose chondroectodermal dysplasia, and refer patients with this syndrome to appropriate health care professionals to receive treatment to prevent further cardiac complications and bone deformities.

Key words: Chondroectodermal dysplasia; Ellis Van Creveld syndrome; Polydactyly

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INTRODUCTION

Ellis-Van Creveld (EVC) syndrome, also called chondroectodermal dysplasia, mesoectodermal dysplasia, or chondrodystrophy, is a rare autosomal recessive congenital disorder [1, 2]. Its reported incidence in Amish population of Pennsylvania, USA, is one in 1,500,000 live births and in non-Amish population, the incidence is seven in 1,000,000 live births [3].

In Iran, however, the incidence of this syndrome has not been previously reported. EVC was first reported by McIntosh in 1933 and was subsequently described in detail by Ellis and Van Creveld in 1940 [4].

EVC (OMIM # 225500), is caused by a mutation in the EVC1 gene or a mutation in a non-homologous gene, EVC2, which is located close to EVC gene in a head-to-head configuration [4, 5]. Parental consanguinity has been observed in about 30% of the cases [1, 5, 6].

This syndrome is characterized by a variable spectrum of clinical findings, among which chondrodystrophy, polydactyly, ectodermal dysplasia, and congenital cardiac anomalies are the most common [5, 7].

About half of the children suffering from EVC syndrome fail to survive because of respiratory problems, and the rest need different procedures to manage their disease [8].

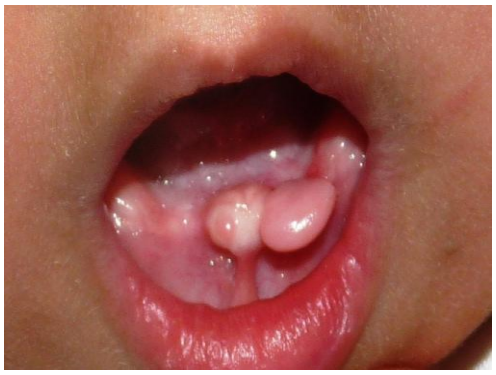

Fig 1. Gingival hypertrophy, labiogingival frenulum hypertrophy, retarded eruption of deciduous teeth, oligodontia and mild serration of alveolar ridge

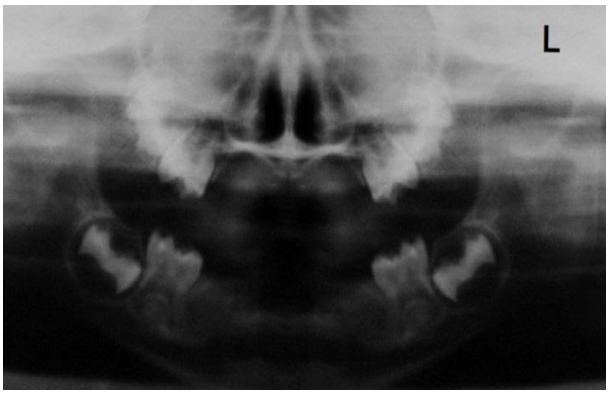

Fig 2. Oligodontia

Fig 3. Polydactyly and dystrophic nails

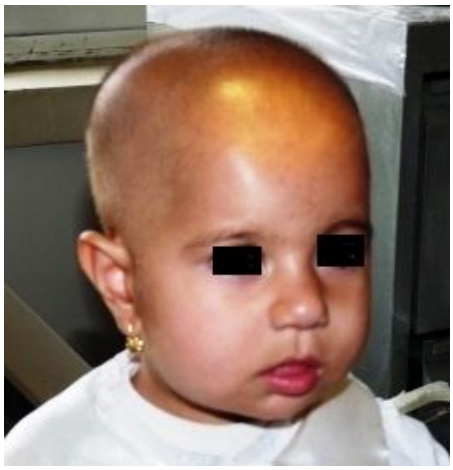

Fig 4. Cutaneous appearance of a two-and-a-half-year-old girl

Oral manifestations of EVC syndrome include hyperplastic frenula, absence of mucobuccal fold, gingival hypertrophy, hypodontia, dystrophic philtrum, and cleft palate [2, 8, 9]. The maxillary and mandibular alveolar processes may present notching or submucosal clefts [8, 9]. Uncommon oral and dental diseases have helped physicians to diagnose this syndrome aptly and refer the patients to potentially prevent additional complications [8].

CASE REPORT

A two-and-a-half year old female presented to the Department of Pediatric Dentistry in Isfahan University of Medical Sciences in April 2012, with the chief complaint of delayed tooth eruption. She was the first child of consanguineous parents. She had no teeth, and exhibited hypertrophy of the labio-gingival frenulum (Figure 1).

All the primary incisors, canines, and first molars, as confirmed by radiographs, were congenitally missing (Figure 2). She did not exhibit gingival hypertrophy, and had missing teeth in the gingival area. The rest of her physical examination was significant for polydactyly (hexadactyly) of both hands, and dystrophic nails (Figure 3). Her head morphology and facial appearance were normal, except for hidrotic and fine hair.

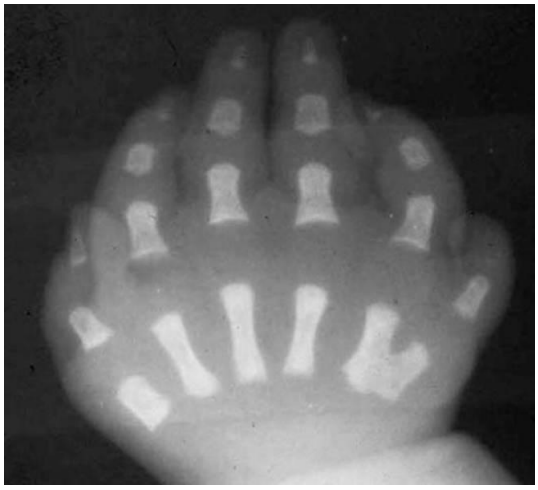

Fig 5. Post-axial polydactyly with fusion of the 5th and 6th metacarpal bones and disproportionately shorter distal phalangeal bones

Her skin appearance was normal, with normal perspiration (Figure 4).

She had post-axial polydactyly with fusion of the Fifth and the Sixth metacarpal bones and disproportionately shorter distal phalangeal bones (acromelia) (Figure 5).

There were no laxity or limitations of movement at any joints. The muscle bulk and tonicity were within normal limits.

She was referred to a pediatric hospital and was examined for anomalies in other organ systems.

Specifically, she was examined for bony abnormalities, as well as defects in her kidneys, heart and lungs. She was reported to have a systolic functional murmur, and shortness of limbs.

DISCUSSION

Ellis-Van Creveld (EVC) syndrome is a rare autosomal recessive congenital disorder [1,2], characterized by chondrodystrophy, polydactyly, ectodermal dysplasia, and congenital cardiac anomalies. Patients may present with growth failure of the proximal tibial epiphysis and the tibial segment may be disproportionately shorter than the femoral segment, or fibula may be shorter than tibia.

Oral manifestations include hyperplastic frenula, absence of mucobuccal fold, gingival hypertrophy, hypodontia, dystrophic philtrum, and cleft palate [2, 8, 9]. The maxillary and mandibular alveolar processes may present notching or submucosal clefts. The differential diagnosis of EVC includes Weyers syndrome, McKusick-Kaufman syndrome and Jeune syndrome [2]. MKK syndrome, which is a recessively inherited disorder, can be distinguished from EVC by its characteristic presence of hydrometrocolpos. In Weyers syndrome, ectodermal disturbances are similar to EVC, but unlike EVC, thoracic dysplasia, congenital heart diseases and dwarfism are absent in Weyers syndrome. Jeune syndrome, an inherited form of dwarfism, is characterized by short limbs, similar to those observed in EVC. However, patients suffering from Jeune syndrome do not exhibit fingernail hypoplasia, which is commonly observed in patients with EVC [8, 10, 11]. Patients with EVC should be managed through a multidisciplinary approach. Particularly during the neonatal period, these patients require treatment for respiratory distress and heart failure. Our patient remained undiagnosed, because she had a functional murmur; which was disregarded. Management of bone deformities requires orthopedic follow-ups and oral manifestations need intense professional attention. Prognosis is coupled to the respiratory problems during the first few months of life that are attributable to thoracic narrowness and heart defects. Treatment should be started as early as possible in order to attain the best possible clinical outcome. Patients may require gingivectomy, frenectomy, and amputation of extra digits under sedation or general anesthesia. Dental care after eruption of teeth includes oral hygiene instructions, dietary counseling and effective plaque control. Children with EVC syndrome are at a high risk for caries. Definitive management throughout adulthood includes dental implants and prosthetic rehabilitation to compensate for congenital oligodontia.

Although EVC syndrome has been reported to be quite rare, our case which was detected under normal conditions can be alarming for dental care providers.

Due to diverse physical problems that such patients may suffer from, and the restrictions they may have for the medications that they take, it is imperative to pay utmost attention while examining or screening these patients for consequent referrals.

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