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Case Report Open Access

Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome): A Case Report with Dental Considerations and Review of Literature

L. Kayal[1]*, S. Jayachandran[2] and Koijam Sashikumar Singh[3]

[1]Reader, Department of Oral Medicine and Radiology, Tamilnadu Government Dental College and Hospital, Chennai, Tamil Nadu, India
[2]Professor and Head, Department of Oral Medicine and Radiology, Tamilnadu Government Dental College and Hospital, Chennai, Tamil Nadu, India
[3]Postgraduate Student, Department of Oral Medicine and Radiology, Tamilnadu Government Dental College and Hospital, Chennai, Tamil Nadu, India

Abstract

Ellis-van Creveld syndrome is a rare autosomal recessive disorder. It is a chondroectodermal dysplasia characterized bychondrodysplasia, polydactyly, ectodermal dysplasia and congenital heart defects. Oral manifestations include fusion of the upper lip to the gingival margin, presence of multiple frenum, abnormally shaped and microdontic teeth, malocclusion, and congenitally missing teeth. Patients with EVC syndrome require multidisciplinary approach. Because of its unique oral and dental manifestations and its association with cardiac problems, dentist plays an important role in managing dental discrepancies and diagnosing possible serious health conditions beforehand.

Keywords: Ellis-van Creveld syndrome; Chondroectodermal dysplasia; Polydactyly

Introduction

Ellis-van Creveld syndrome (EVC) or chondroectodermal dysplasia is a rare autosomal recessive disease caused by mutations of the EVC1 and EVC2 genes positioned in a head-to-head configuration on chromosome 4p16 [1]. It was first described by Richard Ellis and Simon van Creveld in 1940 [1].

The incidence of EvC syndromein general population is low [2]. This syndrome is most prevalent in the Amish population of USA. The birth prevalence in non-Amish population is estimated to be 7/1,000,000 of live birth [3,4].

The principal features of this syndrome are chondroectodermal dysplasia, polydactyly and congenital heart defects. The patients have small stature, short limbs, fine sparse hair and hypoplastic fingernails. Oral manifestations include multiple musculofibrousfrenum, dental transposition, conical teeth, hypoplasia of the enamel, hypodontia and malocclusion. The teeth can erupt and exfoliate prematurely [1].

We describe a clinical case of a patient who visited the Tamil Nadu Govt. Dental College and Hospital, Chennai, Tamil Nadu, India presenting the typical clinical features of EVC syndrome.

Case Report

A 4 and half years old boy was reported to the Department of Oral Medicine and Radiology, Tamil Nadu Govt. Dental College and Hospital, Chennai with complains of multiple missing teeth and caries. The patient was referred from Institute of Child Health, Egmore, Chennai for opinion. There was no history of parental consanguity and the parents were in good health. The patient has an elder sister who is apparently normal. The patient had intelligence in the normal range.

Patient stature was 89 cm, which is relatively short for age. Weight measured 17 kg which was normal. Body mass index (BMI) was measured and it was 21.46. The extremities were plump, and shortness of the limbs was evident (Figure1). Genu vulgam (Figure 1) was seen due to abnormal proximal tibial epiphysis. Fingernails and toenails were markedly hypoplastic, thin and wrinkled (Figure2a, 2b). Bilateral postaxial polydactyly of hands were present (Figure 2a). Hair was sparse especially in the eyebrows (Figure3). The left testis was not fully descended. A family pedigree was analyzedupto three generations and it showed no familial inheritance (Figure 4).

No cardiac abnormality was reported in examination by the cardiologist. Other systems were also normal.

On oral examination, there was a fusion of the middle portion of the upper lip to the maxillary gingival margin (Figure 5). Presence of multiple frenum was seen in the anterior portion of maxillary arch (Figure 5).There were multiple missing deciduous incisors (52,61,71,72,81 and 82) in anterior maxilla and mandible (Figure

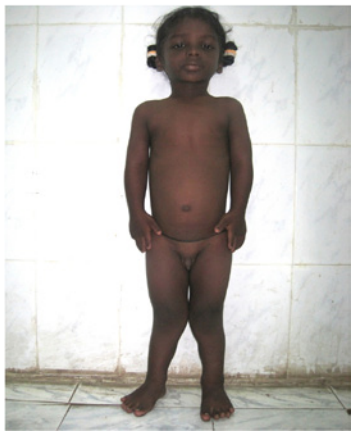

Figure 1:

***Corresponding author:** L. Kayal, Reader, Department of Oral Medicine and Radiology, Tamilnadu Government Dental College and Hospital, Chennai, Tamil Nadu, India, E-mail: drlkayal.kumar@rediffmail.com

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5). There was presence of conical, microdontic teeth (Figure 5). The primary molars have wide grooves and atypical cusps (Figure 6). Multiple caries were present. Enamel hypoplasia was noted on 51,62 and 63.

OPG revealed congenitally missing permanent teeth 12, 21, 31, 32, 41 and 42 (Figure 7). The pulp chambers of the mandibular primary molars had an increased apicoocclusal height and lacked the constriction at the level of the CEJ,resulting in a rectangular shape, and thus resembling the features of taurodontism (Figure 6). Radiograph of the hands revealed bilateral postaxial polydactyly (Figure 8).

Discussion

Ellis-van Creveld syndrome presents a characteristic tetrad [5]:

1. Disproportionate dwarfism due to chondrodysplasia of the long bones and an exceptionally long trunk. The severity of short limbs increases from the proximal to the distal portions.
2. Bilateral postaxial polydactyly of the hands, with the supernumerary finger usually being on the ulnar side. Occasionally a supernumerary toe may be present.
3. Ectodermal dysplasia with dystrophic, small nails, thin sparse hair, and hypodontic and abnormally formed teeth.
4. Congenital heart malformations in 50% to 60% of cases, the most common being a single atrium and a ventricular septal defect.

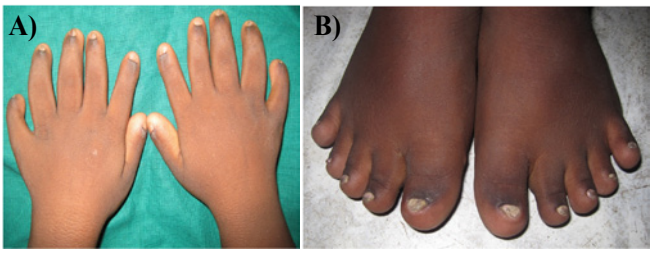
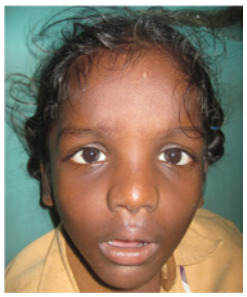


Figure 2:



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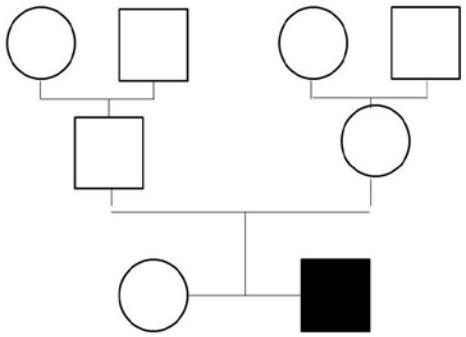

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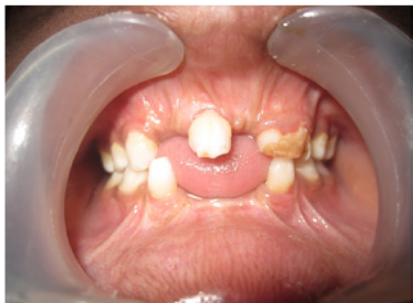

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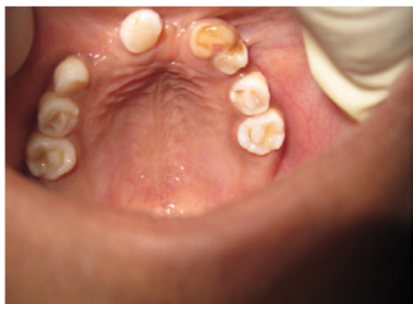

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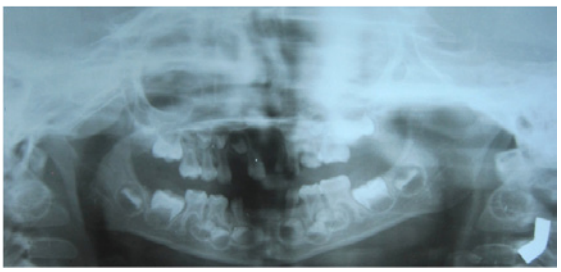

Figure 7:

Other skeletal anomalies such as genu valgum have occasionally been reported [6]. Patient's intelligence is usually normal [7].

Normal height of a 4year-old boy is 102.9 cm and 109.9 cm for a 5-year-old boy [8]. The height of the patient was only 89 cm which was shorter than normal. There were no cardiac abnormalities associated with the patient in this case. The finding that the left testis was not fully descended seen in the patient is an association not reported so far. Identification of this finding in other patients with EvC syndrome is necessary to validate its association with the syndrome.

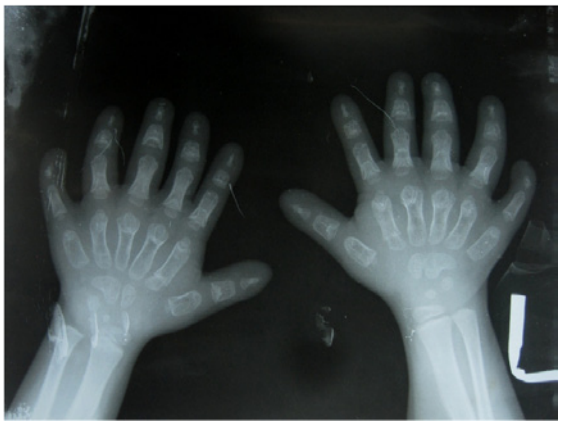

Figure 8:

Prenatal diagnosis can be made with intrauterine growth retardation, skeletal malformations and cardiac defects on ultrasound images. Diagnosis is also possible using chorionic villi or amniotic fluid using linked-microsatellite markers if a previously affected sibling has been identified [9].The prognosis of EvCsyndrome is linked to the presence of congenital heart defects and respiratory difficulties in the first months of life. Patients without cardiac or respiratory problems may live a normal life span as that of a normal individual.

Oral manifestations include fusion of the upper lip to the gingival margin, presence of multiple frenum, conical, microdontic teeth, molars with wide grooves and atypical cusps, congenitally missing teeth [7]. Taurodontism, a feature that has been reported in permanent and some primary molars [7], was found in primary mandibular molars in the patient.

Dental management includes fabrication of crowns for microdontic and conical teeth, modification of abnormal grooves and cusp to avoid early caries and achieve occlusal harmony, topical and systemic fluoride supplements to reduce the incidence of caries, fabrication of missing teeth with temporary prosthesis during the growing phase and with permanent counterpart when growth ceases. Frenectomy and vestibuloplasty may also be considered in case of difficulty in speech and retention of dental prosthesis.

EvC syndrome involves all the embryonic tissue layers and is polysymptomatic [10]. A multidisciplinary approach is encouraged for proper management and rehabilitation of patients with EvCsyndrome. The dentist must bear in mind the high prevalence of cardiac defects in these patients and implement established procedures for the prevention of bacterial endocarditis when necessary. Dental treatment should be made under low antibiotic prophylaxis.

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