

Case Report

Ellis–van Creveld syndrome with unusual oral and dental findings: A rare clinical entity

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ABSTRACT

Ellis–van Creveld (EVC) syndrome, a form of skeletal and chondroectodermal dysplasia, is an autosomal recessive disorder characterized by a tetrad of disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, and heart defects. In the present article, we hereby present a case of a 13-year-old girl of Indian ethnicity with EVC syndrome with a remarkable number of classical oral and dental features, with unusual findings such as taurodontism and talons cusp. Such dental findings were reported in few cases only. Despite the fact that oral manifestations play an important role in the diagnosis of EVC, only a few detailed reports have been published in the dental literature.

Key Words: Ellis–van Creveld syndrome, polydactyly, taurodontism

INTRODUCTION

Ellis–van Creveld syndrome (EVC) or Chondroectodermal dysplasia is a rare disease complex, where all the three embryonic layers appear to be involved.^[1] The first full description of the syndrome was given by Ellis and van Creveld in 1940.^[2] The exact prevalence remains unknown; even so the birth prevalence of EVC has been estimated to be 7 per 1,000,000 population. It is caused by mutations of the EVC1 and EVC2 genes and shows autosomal recessive inheritance with parental consanguinity in about 30% of cases.^[3] EVC is found with an increased frequency among the Amish community in Lancaster county, Pennsylvania, US, where the largest pedigree has been described 52 cases in 30 sibships.^[4] Very few cases have been reported among Indian population.

The EVC phenotype is variable and affects multiple organs. Skeletal dysplasia is present at birth, being

characterized by short limbs, especially the middle and distal segments, accompanied by postaxial polydactyly of the hands and sometimes the feet. Nearly, all patients show severe dystrophy of the fingernails, which are markedly hypoplastic, thin, and often spoon-shaped. Congenital heart malformations are present in about 50–60% of cases.^[1,5]

EVC shows a wide spectrum of oral manifestations, including malocclusion, labiogingival adhesions, labiogingival frenulum hypertrophy, accessory labiogingival frenula, serrated appearance of the gingiva, dental transposition, diastema, conical teeth, enamel hypoplasia, and hypodontia. Teeth may show premature eruption at birth or premature exfoliation. Supernumerary teeth may also be present.^[5,6]

We hereby present a case report of a female child of Indian ethnicity manifesting many of the classical

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features of EVC, but with unusual dental findings such as talons cusp and taurodontism. According to the previous literature, there is only one case report describing the association between EVC and talon's cusp and two cases reporting about taurodontism.

CASE REPORT

A 13-year-old girl of Indian origin reported to our institution with the chief complaint of malformed teeth in the front region of upper and lower jaws. The patient was the only child of consanguineous and normally developed parents and was born 10 years after marriage. There was no history of the presence of natal or neonatal teeth.

General examination revealed that the patient had disproportionate short stature. The limbs were short with postaxial polydactyly affecting both hands [Figure 1]. Other findings were hypertelorism, broad depressed nasal bridge, and short bulbous nose. Other striking features included shortening of middle and distal phalanges, wide space between hallux and the rest of phalanges, wide space between hallux and the rest of

the toes, and short 2nd toe of right foot overlapping over the 3rd toe [Figure 2]. Her nails were dystrophic, friable, markedly hypoplastic, and thin [Figures 1 and 2].

Intraoral examination revealed fusion of the middle portion of the upper lip to the maxillary gingival mucosal margin, and lower lip to the mandibular gingival mucosal margin with the absence of the mucobuccal fold, labiolingual adhesences, multiple abnormal frenula [Figures 3 and 4], serrations of the alveolar ridge were observed distally to the upper central incisors [Figure 5], obliteration of both buccal and labial vestibule, missing teeth irt 12, 22, 31, 41, 42, conical tooth irt 32 with labiolingual rotation, decayed tooth irt 36, and Talon's cusp irt 21 [Figure 6].

Panoramic radiograph revealed missing upper right and left lateral incisors and mandibular central incisors; taurodontism irt 16, 17, 26, 27; conical crown

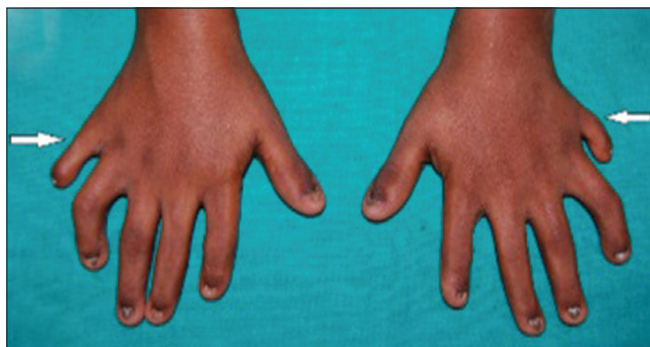


Figure 1: Postaxial polydactyly in both hands (white arrows).



Figure 2: Wide space between the great toe and the rest of the toes (white arrows).



Figure 3: Multiple abnormal frenum in maxillary anterior sulcus (white arrows).



Figure 4: Multiple abnormal frenum in mandibular anterior sulcus (white arrows).

with short roots irt 32; impacted tooth with conical crowns and short root irt 42; radiolucency involving enamel-dentin and pulp irt 36 with horizontal and vertical bone loss was seen [Figure 7].

Lateral skull radiograph revealed normal anatomic structures, except for maxillary prognathism [Figure 8]. Hands and wrist radiography revealed polydactyly [Figure 9].

The patient was further subjected to echocardiography which revealed no evidence of congenital heart defects [Figure 10]. The patient's intellectual ability was within the normal range.

Based on the clinical and radiographic findings of the dental and subsequent medical examinations, the medical team diagnosed the patient as having EVC syndrome. Written informed consent was obtained from the patient's parents to report the case.

Surgical removal of impacted 42 was done along with the extraction of 32 and grossly decayed 36. Further,

the patient was advised full mouth rehabilitation; however, the patient was unwilling for further treatments due to personal reasons.

DISCUSSION

EVC is an autosomal recessive disorder. In the present case, the patient had a history of consanguineous marriage of parents, which supports the recessive nature of the disorder.

The patient, living in a rural area, had not undergone any comprehensive examinations before being referred to the Department of Oral Medicine and Radiology, where oral findings in addition to other clinical features led us to suspect EVC syndrome. The present patient showed all of the typical clinical and radiographic oral features of EVC.

Aminabadi^[7] reported that fusion of the upper lip with the gingival margin as observed in our case is characteristic of this syndrome which could be due to retardation of



Figure 5: Clinically missing maxillary left and right lateral incisors and serrations of the alveolar ridge (white arrows).



Figure 6: Talon cusp irt 21 (white arrow).

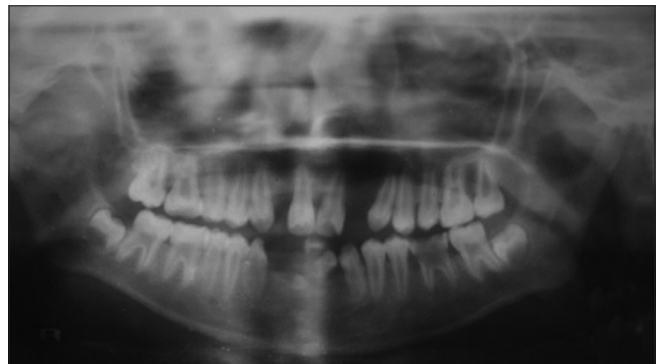


Figure 7: Panoramic radiograph revealing missing upper right and left lateral incisors and mandibular central incisors; taurodontism irt 16, 17, 26, 27; conical crown with short roots irt 32; impacted tooth with conical crowns and short root irt 42.



Figure 8: Lateral skull revealing maxillary prognathism.

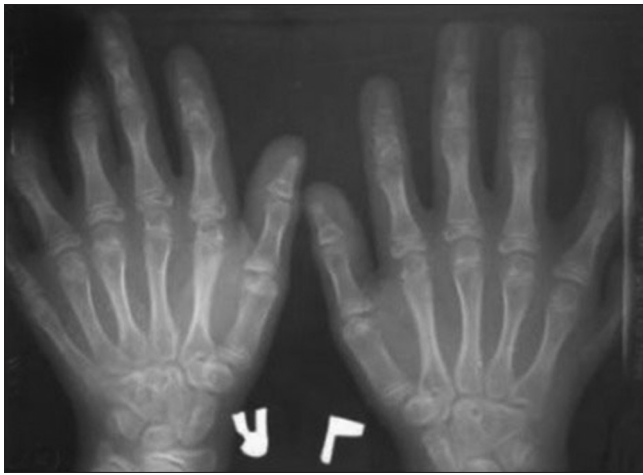


Figure 9: Radiograph of hands and wrist showed polydactyly.



Figure 10: Echocardiogram revealing no abnormality.

bone development in the alveolar ridge, as is known to occur with other bones. Small alveolar notches on the crest of the thin alveolar ridge were evident, involving the areas normally occupied by the upper lateral incisor and such similar finding was observed by Biggerstaff and Mazaheri.^[8] Each submucousal cleft was marked by a moderately sized fibrous band, whose fibers appeared to incise the underlying alveolar process and extend across the mucobuccal fold into the lip. Between the two incomplete clefts and the moderately sized fibrous bands were numerous smaller fibrous bands which, when considered together, formed a continuous frenulum that reduced the extent of the attached gingiva and the labial sulcus. According to Gorlin *et al.*,^[9] this notching is a continuation of the normal serrated condition, which is present from the 3rd to the 7th month *in utero*.

In this patient, four permanent teeth, other than the third molars, were absent. Such severe hypodontia

beyond the incisor region has also been reported by Hunter and Roberts.^[6]

In patients with EVC syndrome, the teeth to erupt tend to be small, conically crowned, or resembling bicuspid with an accentuated cusp height, and deep steep-sided fissure patterns. This may explain the high incidence of caries in affected patients,^[6] although enamel hypoplasia might be another reason. Taurodontism, a feature that has been reported previously^[6,7] in permanent and some primary molars was found in upper permanent molars in the present patient. However, present case exhibited unusual finding, talons cusp, which was reported in only one case in previous literature by Hattab *et al.*^[10]

Management of EVC requires a multidisciplinary approach which involves oral physician, oral radiologist, cardiologist, ophthalmologist, psychologist, and clinical geneticist.

Dental counseling, plaque control, and oral hygiene maintenance are important in the prevention of caries, short conical teeth can be corrected by composite or fixed crowns. Space maintainers can be advised to replace congenitally missing teeth in the form of partial dentures for the improvement of aesthetics, speech, and mastication. Orthodontic intervention is required for the correction of dental malocclusion.

Extraction of unerupted and horizontally impacted 42 was performed as there was no chance of this tooth to erupt into oral cavity. Approximately, 32 were extracted since it had short conical crown and was mobile, and 36 was grossly decayed tooth. No further dental treatments could be performed in this case due to patient's personal reasons.

CONCLUSION

We hereby report rare and unusual dental findings such as talon's cusp and taurodontism associated with EVC syndrome. Dentists play an important role in the diagnosis and the management of oral and dental manifestations associated with this syndrome.

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Conflicts of interest

The authors of this manuscript declare that they have no conflicts of interest, real or perceived, financial or non-financial in this article.

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