Case Report

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# Chondroectodermal Dysplasia without Cardiac Anomalies: A Rare Case Report

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## ABSTRACT

**Background**: Chondroectodermal dysplasia is a rare autosomal recessive disorder with a complex phenotype characterized by chondrodystrophy, postaxial polydactyly, ectodermal dysplasia and cardiac anomalies. We present the case of a 19 year-old Indian male with unaffected parents and an affected elder sister. No cardiac anomalies were noticed and bilateral post axial polydactyly was evident in both the upper and lower limbs. The importance of oral care should be emphasized in affected patients without cardiac abnormalities as they have increased life expectancy and oral rehabilitation could improve the quality of life in these patients.

**Keywords:** Postaxial polydactyly; ectodermal dysplasia; chondrodystrophy; Ellis-van creveld syndrome.

## INTRODUCTION

Chondroectodermal dysplasia (CED) is a rare type of skeletal dysplasia classified as a type of mesomelic limb shortening.[1] It is an autosomal recessive disorder caused by mutations in two non-homologous genes, EVC and EVC2, located in a head to head configuration on chromosome 4p16.[2,3] Whilst no gender predilection has been reported, the incidence in general population has been reported to be around 1 per 60000 live births.[3] CED is characterized by a tetrad of chondrodystrophy, hidrotic ectodermal dysplasia, post-axial polydactyly, and congenital heart defects. This article presents a rare case of CED affecting a 19 year-old male who had presented without cardiac anomalies and with bilateral post axial polydactyly affecting both the upper and lower limbs.

## CASE HISTORY

A 19 year-old Indian male reported with the chief complaint of missing upper and lower front teeth. Family history revealed that he was the third child of consanguineously married healthy parents. Detailed anamnesis revealed that pregnancy and childbirth were uneventful. His medical history was non-contributory and developmental history revealed that the patient started walking at the age of three years and the teeth started erupting at the same age.

The patient was short statured and appeared relatively shorter for his age. Whilst his head morphology, facial appearance, quantity and quality of the hair were normal, skeletal deformities noticed included bimanual postaxial polydactyly, shortened and markedly deformed limbs (genu valgum), short middle and distal phalanges, and dystrophic finger and toe nails. **[Figure 1]** Parents reported that his elder sister [second child] shared the same phenotypic features as the proband. Intraoral manifestations were remarkable and showed edentulous mandibular incisor region with serrated alveolar ridge, mandibular hyperplastic frenulae with absence of anterior mucobuccal fold.

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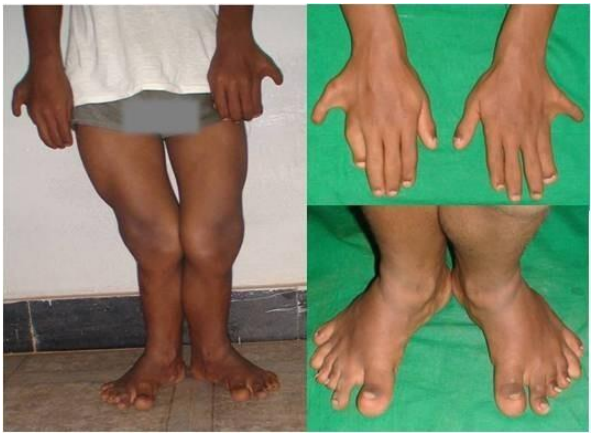

Fig 1: Picture showing evidence of short stature, genu valgum, bimanual postaxial polydactyly and dystrophic nails.

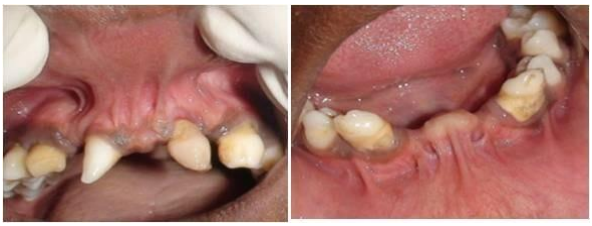

Fig 2: Intraoral picture showing labiogingival frenulum hypertrophy, accessory frenula, serrated mandibular anterior alveolar ridge, oligodontia and malformed teeth.

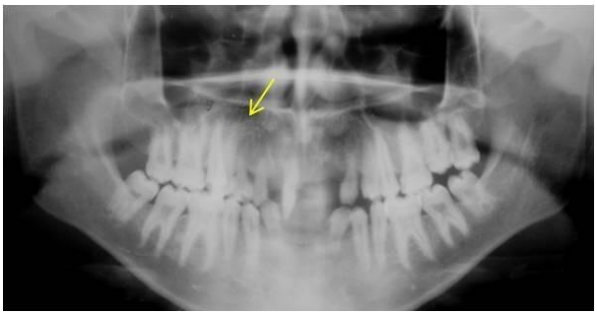

Fig 3: OPG showing supernumerary tooth in the maxillary right premolar region.

The labial frenum of the upper lip adhered to the anterior alveolar ridge obliterating the mucolabial sulcus. Maxillary permanent lateral incisors were missing on both sides. The erupted maxillary anteriors and the permanent molars of both arches were malformed. **[Figure 2]** A supernumerary tooth was evident in the premolar region of the first quadrant. There was no history of either natal/neonatal teeth and of previous exfoliation/extraction of incisors.

Panoramic radiograph revealed congenitally missing 12, 22, 31, 32, 41 and 42 [FDI] and a supernumerary tooth in the maxillary right premolar region. **[Figure 3]** A hand-wrist radiograph revealed shortening of the middle and distal phalanges, post-axial polydactyly, fusion of hamate and capitate bones, and fusion of $5^{th}$ and $6^{th}$ metacarpal bones. **[Figure 4]** The patient was referred for medical examination which revealed no evidence of systemic involvement including cardiac abnormalities. The patient's psychomotor and cognitive developments were within normal limits. Based on the clinical and radiographic findings and subsequent medical examinations, the patient was diagnosed as having chondroectodermal dysplasia.

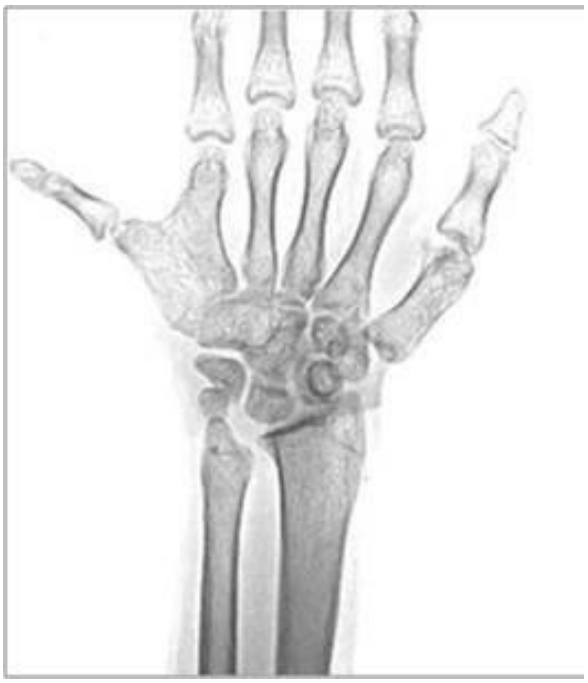

Fig 4: Hand-wrist radiograph showing fusion of hamate and capitate bones, and fusion of the 5th and 6th metacarpal bones (left hand).

Treatment plan included mucogingival surgery for excision of the labial frenulum in both upper and lower arches, followed by prosthetic rehabilitation of his missing teeth. Extraction of the supernumerary tooth, restorative care for malformed teeth, and home care oral hygiene measures were advised. Unfortunately, the patient failed to return for further treatment.

## DISCUSSION

Chondroectodermal dysplasia (CED) or Ellis-van creveld syndrome is characterized by multiple abnormalities of the mesodermal and ectodermal tissue. Whilst no specific constant features in CED have been described in the literature, the characteristic clinical tetrad of this condition includes chondrodystrophy resulting in shortening of long bones, hidrotic ectodermal dysplasia affecting the nails, hair and teeth, post-axial polydactyly, and congenital heart defects. Radiological skeletal features include fusion of the hamate and capitate bones of the wrist, knock-knees, fusion of the 5th and 6th metacarpals and retarded bone maturation.[3]

Diverse spectrum of oral manifestations are seen in this syndrome including oligodontia, enamel hypoplasia, dental transposition, conical or malformed teeth, malocclusion, labiogingival frenulum hypertrophy, accessory labiogingival frenula, and serrated appearance of the lower anterior alveolar ridge.[3,4] Mostafa et al (2005)[5] had suggested that midline orodental anomalies are present only in cases of CED. The findings seen in our patient are in accordance with the typical features present in CED, as reported in the literature. Congenital heart malformations are described in 50 to 60% of patients affected by this syndrome and is the principal cause of decreased life-expectancy in these patients.[3,6,7] Further, patients who survive infancy have increased chances of normal life expectancy as seen in our patient who showed no evidence of cardiac anomalies which could have significantly improved his survival years.

Polydactyly is usually bilateral and affects the hands while the feet are less affected.[3,4] However in our case, both the limbs were affected. The presence of supernumerary tooth in our case agrees with the findings of other authors.[4,8] Orofacialdigital syndrome was considered in the differential diagnosis. However, absence of mental retardation, nasal cartilage hypoplasia, fissured tongue and/or ankyloglossia and dominant sex-linked inheritance excluded it from the final diagnosis.[9]

## CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

## CONCLUSION

CED requires multidisciplinary treatment planning and the dentist plays a major role in the management of the oro-dental manifestations as they constitute one of the characteristic diagnostic features. The importance of oral care should be emphasized in CED patients without cardiac abnormalities as they have increased life expectancy and oral rehabilitation could improve the quality of life in such patients.

## REFERENCES

1. Nyberg DA, Mc Gahan JP, Pretorius DH. Diagnostic imaging of fetal anomalies. Lippincott Williams & Wilkins. 2003. ISBN: 0781732115.

2. Ruiz-Perez VL, Tompson SWJ, Blair HJ et al., Mutations in two nonhomologous genes in a head-to-head configuration cause Ellis–van Creveld syndrome. Am J Hum Genet 2003;72:728–32.

3. Baujat G, Le Merrer M. Ellis-van Creveld syndrome. Orphanet J Rare Dis 2007;2:27.

4. Kurian K, Shanmugam S, Harsh Vardah T, Gupta S. Chondroectodermal dysplasia (Ellis van Creveld syndrome): a report of three cases with review of literature. Indian J Dent Res 2007;18:31-34.

5. Mostafa MI, Temtamy SA, El-gammal MA, Mazen IM. Unusual pattern of inheritance and orodental changes in the Ellis–van Creveld syndrome, Genet Counsel 2005;16:75–83.

6. Katsouras CS, Thomadakis C, Michalis LK. Cardiac Ellis-van Creveld syndrome. Int J Cardiol 2003;87:315-16.

7. Arya L, Mendiratta V, Sharma RC, Solanki RS. Ellis-van Creveld Syndrome: a report of two cases. Pediatr Dermatol 2001;18:485-9.

8. Aminabadi NA, Ebrahimi A, Oskouei SG. Chondroectodermal dysplasia (Ellis-van Creveld syndrome): a case report. Journal of Oral Science 2010;52:333-36.

9. Ghosh S, Setty S, Sivakumar A, Pai KM. Report of a new syndrome:focus on differential

diagnosis and review of Ellis-van Creveld, Curry-Hall, acrofacial dysostosis, and orofacial digital syndromes. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2007;103:670-6.