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Case Report

A perfect diagnosis of Ellis-van Creveld syndrome by oral manifestations: a case report

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ABSTRACT

Ellis-van Creveld syndrome is an extremely rare congenital genetic disorder having autosomal recessive inheritance. The characteristic features of this syndrome are bilateral postaxial polydactyly, acromesomelic dwarfism, ectodermal dysplasia affecting nails, congenital cardiac malformation, edentulous maxillary and mandibular incisors, non-appearance of mucobuccal fold, congenitally missing teeth, slight serrations of the alveolar ridge and multiple small alveolar notches. The present case describes the oral manifestations of the patient which leads to a perfect diagnosis of this syndrome. Ellis-van Creveld syndrome requires a multidisciplinary management and, hence the dental surgeons play an important role in these cases.

Keywords: Chondroectodermal dysplasia, Ellis vancreveld syndrome, Oral manifestations

INTRODUCTION

Ellis-van Creveld syndrome is an extremely rare congenital genetic disorder having autosomal recessive inheritance. It was first discovered by Richard W.B. Ellis and Simon van Creveld in 1940. It is also known as chondroectodermal dysplasia or mesoectodermal dysplasia. Chondroectodermal is the term used to describe the types of tissues involved in the disorder, mainly involve the long bones of the skeleton, nails and teeth. Mesoectodermal dysplasia is the term once proposed to include the 60% incidence of congenital heart disease that associate with the disorder.[1] Mutations of the EVC1 and EVC2 genes are in a head to head configuration on chromosome 4p16, which is the causative factor.[2] This report demonstrates a typical Ellis vancreveld syndrome in a 14-year-old Indian girl with the classical oral manifestations, which will aid the dental surgeons in diagnosing the syndrome and refer to other health care professionals to prevent from further complications.

CASE REPORT

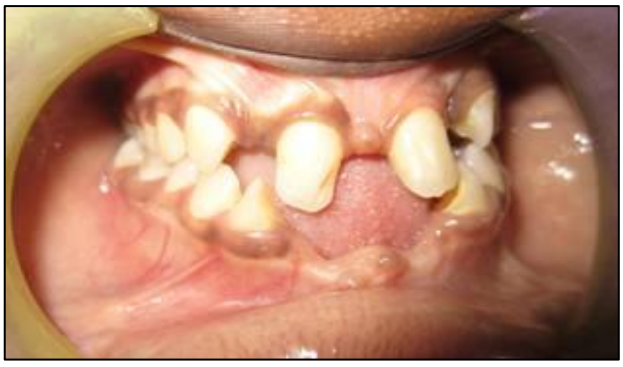

Figure 1: Missing lower incisors and upper laterals.

A 14 year old female patient reported to our dental outpatient department with the chief complaint of missing teeth in relation to upper and lower anterior region (Figure 1). Medical history revealed that the patient had a congenital heart defect and bilateral genu valgum, corrected before 3 years. Familial history revealed that she was the only child of consanguineously married and normally developed parents. Pregnancy and birth was uneventful. The girl was of short stature, upper and lower limbs were short, fingernails and toe nails were hypoplastic, thin and spoon-shaped, bimanual hexadactyly and polysyndactyly of toes are present (Figure 2, 3, 4).

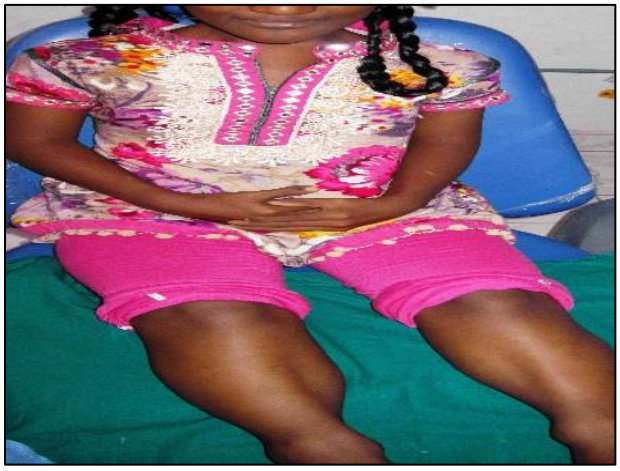

Figure 2: Shortened upper and lower limbs.

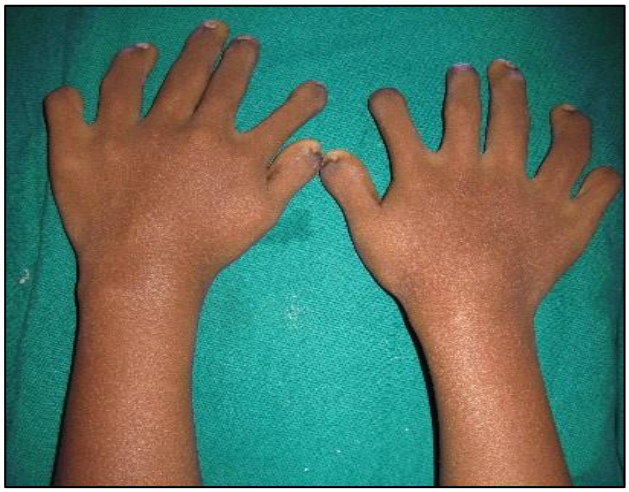

Figure 3: Polydactyly with hypoplastic and spoon shaped nails.

Facial appearance of the girl was leptoprosopic. Intraoral examination revealed edentulous mandibular incisors, absence of maxillary laterals, midline diastema, absence of mandibular anterior mucobuccal fold, presence of mandibular hyperplastic freenum, morphological abnormalities of maxillary and mandibular anterior teeth and slight serrations of the upper alveolar ridge were there (Figure 5, 6). Panoramic radiograph showed, missing permanent mandibular incisors, delayed formation of tooth buds of permanent maxillary right and left lateral incisors (Figure 7).

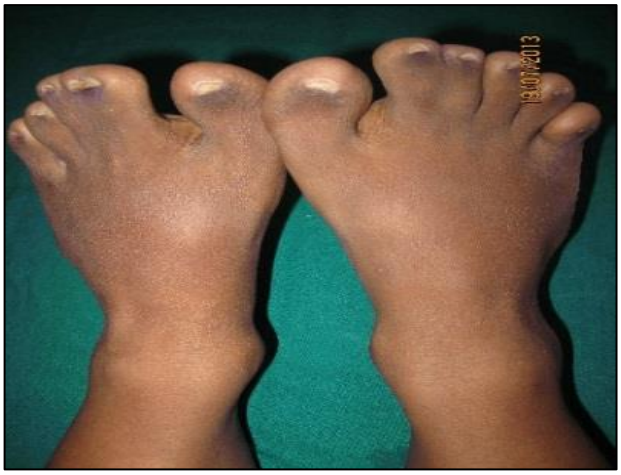

Figure 4: Polysyndactyly.

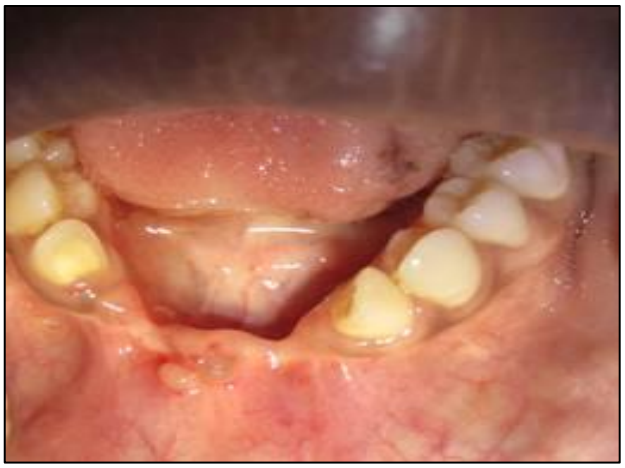

Figure 5: Hyperplastic lower labial fraenum.

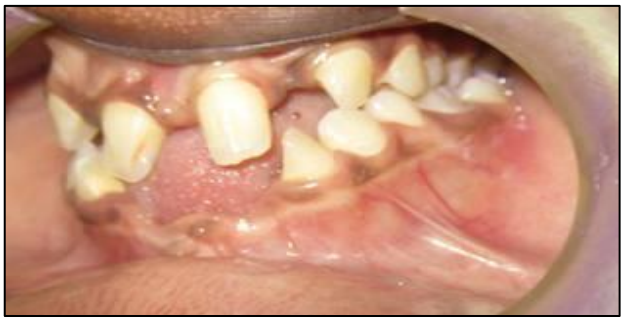

Figure 6: Morphologic abnormalities of teeth.

Radiograph of the upper and lower limb showed shortening of the extremities, polydactyly and polysyndactyly. Based on the clinical and radiographic findings, a final diagnosis of Ellis-van Creveld syndrome was given. An implant supported fixed partial denture was given from canine to canine in the lower anterior

region and the upper central incisors had a root canal treatment with a fixed partial denture from canine to canine.

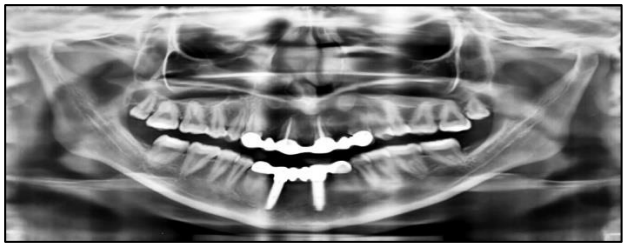

Figure 7: Panoramic view.

DISCUSSION

Ellis-van Creveld syndrome occurs in 1 in 60,000 to 200,000 new-borns. It is difficult to estimate the prevalence because the syndrome is very rare in the general population.[3] This condition is more common in the Amish population of Lancaster County, Pennsylvania.[4] Mutations of the EVC1 and EVC2 genes are in a head to head configuration on chromosome 4p16 which is the causative factor. The characteristic features of the syndrome are bilateral postaxial polydactyly of the hands, acromesomelic dwarfism, ectodermal dysplasia affecting nails as well as teeth and congenital heart malformation.[5] All of these features were present in our case. One more characteristic feature of this syndrome is the familial history which may include parental consanguinity or affected siblings or family members 6, which correlates with our case. Our case was the child of consanguineously married and normally developed parents.

Oral manifestations of the syndrome include the non-appearance of mucobuccal fold in the anterior region, notching of the alveolar ridge, congenitally missing mandibular incisors, erupted teeth having small crowns, irregular spaces between teeth, delayed formation of tooth buds.[7] All these features are consistent with our case. Mostafa et al reported a new consistent orodental anomaly (bifid tip of the tongue) in six Egyptian cases of Ellis-van Creveld syndrome. However, this anomaly was not reported in our patient.[8]

The differential diagnosis of Ellis-van Creveld syndrome is Weyer's syndrome, asphyxiating thoracic dystrophy and orofacial digital syndrome. The features of Ellis-van Creveld syndrome overlap with Weyers acrofacial dysostosis. Similar to Ellis-van Creveld syndrome, Weyers acrofacial dysostosis involves tooth and nail abnormalities, affected people have less pronounced short stature, delayed fusion of mandibular symphysis and do not have any heart defects. The mutations in the same genes have caused these two conditions.[9]

Patients with Ellis-van Creveld syndrome and asphyxiating thoracic dystrophy have similar features in hand, pelvis, and long bones. Hence, it is not possible to differentiate these two conditions radiographically. Cardiac anomalies, hypoplasia of nails, fusion of upper lip to gingiva and neonatal teeth are present in Ellis-van Creveld syndrome. Renal failure with hypertension are present in asphyxiating thoracic dystrophy which will help in distinguishing these two disorders.[10]

Presence of multiple gingivolabial freenum is similar to both Ellis-van Creveld syndrome and orofacial digital syndrome. Ankyloglossia, moderate mental retardation, Hypoplastic nasal cartilage and fissured tongue helps to differentiate orofacial digital syndrome from Ellis-van Creveld syndrome.[10]

CONCLUSION

Genetic counselling is needed to make the parents aware of the risk of recurrences. The diagnoses of Ellis-van Creveld syndrome are depending upon clinical and radiological evaluation. Oral manifestation is one of the characteristic diagnostic features; dentists are having a key role to play in proper management of these cases. The successful management of this syndrome will need a team which includes Paediatric dentist, oral and maxillofacial surgeon, prosthodontist, geneticist, cardiologist, orthopaedician, urologist, psychologist and paediatrician.

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