Case Report

Ellis-Van Creveld syndrome – a case report

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Abstract
Ellis-van Creveld (EVC) syndrome or chondroectodermal dysplasia is a rare autosomal recessive disorder characterized by a variable spectrum of clinical findings. Classical EVC syndrome comprises a tetrad of clinical manifestations of chondrodystrophy, polydactyly, ectodermal dysplasia and cardiac defects. This syndrome manifests with several skeletal anomalies, oral mucosal and dental anomalies, congenital cardiac defects and nail dysplasia.

The presence of oral mucosal and dental abnormalities like notching of alveolar process, fusion of upper lip with gingival margin, oligodontia and conical shape of anterior teeth will confirm the diagnosis of Ellis van Creveld syndrome and hence its importance to dentist.

Keywords:
Polydactyly, Chondroectodermal dysplasia, Skeletal anomalies

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Introduction

Chondroectodermal dysplasia is a complex genetic syndrome first described in 1940 by Ellis and van Creveld (1940) which is also called mesoectodermal dysplasia, Ellis-van Creveld (EVC) syndrome or chondrodystrophy syndrome. A large number of cases were reported in the Amish community of Lancaster, Pennsylvania, USA by McKusick in 19641. Today, this syndrome has been described in other population and it is known to affect all races. It is a rare autosomal recessive disorder whose minimal diagnostic criteria include postaxial polydactyly of the hands, short limb dwarfism and dysplastic fingernails and teeth2. A case is described with some remarkable general and oral manifestations which required orthodontic treatment.

Case report

A 19 years old female patient complained of absence of lower front teeth since 6 months of age with deformity of her fingers of hands and legs since birth.

Detailed history from her parents revealed that the lower midline teeth have not erupted when the child was 6 months of age. By three years of age all of her teeth had erupted except the lower front teeth. They also revealed that they noted the deformity in the fingers of her hands and feet since birth. The patient consulted a dentist at 15 years of age and the patient was referred to the OPD for further evaluation. Personal and family history of the patient revealed that both the parents are alive and their marriage relationship shows a first degree of consanguinity.

General examination of the patient revealed that the patient is calm, cooperative, conscious, and well oriented. The patient was well nourished and moderately built.

Extra oral examination of the patient revealed that the height of the child is less when compared to her age. The hair appears to be normal. She has facial deformity with hypoplasia of the middle thirds of the face with short and centrally depressed upper lip and lower lip. She has small, thick and plumpy hands. The hands have normal joint contours with a hypermobile elbow joints. She had hexadactyly on her left and right hand on the ulnar side of the respective hand (figure 1). The nails of the fingers of both the hands appear atrophied. She has presence of genu valgum with gross calcaneous valgus (figure 2, 3). The length of the femur appears normal where as the tibia and fibula appears to be shortened. The nails of the toe appear atrophied.

Intraoral examination revealed the presence of only 23 teeth with 23, 42,41,31,32 missing. The gingiva is pale pink in colour with normal scalloping seen in relation to all teeth except the upper and lower anterior teeth (figure 4). The middle third of the upper and lower lip is defective and gets attached to the abnormal labial frenum resulting in the obliteration of the upper and lower labial sulcus. Alveolar notching is seen in relation to the maxillary and mandibular anterior region as a small elevated, hard nodule on the alveolar ridge.

Considering the above features a differential diagnosis of the following was made:
Ellis van Creveld syndrome or Chondro-ectodermal dysplasia
- Ectodermal dysplasia
- Achondroplasia

OPG reveals congenitally missing tooth in relation to 23, 32, 31, 41, 42 (figure 5). Hand AP View showing the inferoradioulnar joint which is normal with presence of hexadactyly in the left and right hands (figure 6). PA skull revealing presence of a bosselated frontal air sinus seen in the midline which is symmetrical on both sides with white opaque septae traversing the sinus vertically (figure 7). There is distinct haziness of left maxillary sinus and pneumatization of the mastoid air cells are seen bilaterally. Considering all the above features a final diagnosis of Ellis Van Creveld Syndrome or chondroectodermal dysplasia was given. Parents were made aware of the occurrence of possible future complications in the patient, and advised periodic review with a general practitioner and cardiologist. Regarding the absence of teeth the patient was given a removable partial denture in relation to 32 31 41 42 and was asked to review periodically. Patient was referred to orthodontics department for fixed appliance therapy for malformed teeth. The prosthodontist were later planning for a fixed partial denture in relation to 42 41 31 32.

Figure 1: Hands Showing Polydactyly

Figure 2: Genu Valgum

Figure 3: Genu Valgum

Figure 4: Intraoral Photograph
**Discussion**

EVC phenotype is variable and affects multiple organs. Prenatal abnormalities may be early discovered, after the 18th gestation week; they include narrow thorax, marked shortening of the long bones, hexadactyly of hands and feet and cardiac defect.\(^2,3\) However after birth, the cardinal features usually present are: (1) disproportionate small stature and shortening of the middle and distal phalanges; (2) polydactyly affecting hands; (3) ectodermal dysplasia mainly affecting nails and teeth; (4) congenital heart malformation\(^1,4,5\). In the case reported here, we found all the tetrad of principal features of EVC syndrome.

Oral manifestations in EVC are diverse and include; labiogingival adherences, multiple frenula, sub mucosal clefts, congenitally missing teeth, conical and microdontic teeth, enamel hypoplasia\(^3, 5, 6\). All the above-mentioned abnormalities were present in our case.

A variety of skeletal features observed in our case, have also been reported in
previous case reports. These include; Acromesomelia (relative shortening of the distal and middle segment of the limbs), bilateral postaxial polydactyly, genu valgum (knock- knees), long and narrow thorax. Congenital cardiac malformations occur in about 50-60% of cases and mostly they are in the nature of inter- septal defects.

Ellis van Creveld syndrome is a rare autosomal disorder. One third of these patients die of cardiac or respiratory distress in infancy. Prenatal diagnosis in regard to intrauterine growth retardation, skeletal malformations and cardiac defects can be depicted on ultrasound images. Diagnosis is also positive using chorionic villi or amniotic fluid using linked- microsatellite markers if a previously affected sibling have been identified. Oral findings were primarily responsible in leading to diagnosis of this syndrome.

Conclusion

A multidisciplinary approach is advocated involving a clinical geneticist, cardiologist, pulmonologist, orthopedician, physical and occupational therapist, dentist, pediatrician and pediatric neurologist for proper management and rehabilitation of such cases. The dentist plays an important role in control of dental and oral manifestations. The dental treatment must be performed under prophylactic antibiotic coverage with consideration for cardiac defects in such patients.

References


