Ellis–van Creveld

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Received : 01-12-11
Review completed : 02-01-12
Accepted : 26-01-12

ABSTRACT

Ellis–van Creveld (EVC) syndrome is an autosomal recessive disorder that is also known as chondro-ectodermal dysplasia. The common manifestations of this syndrome are short ribs, postaxial polydactyly, growth retardation, and ectodermal and cardiac defects. The present case report is about an 8-year-old boy who had the features of bilateral hexadactyly, knocked knees, cardiac problems, congenital absence of incisors, fused upper and lower labial frenulum, and mulberry molars.

KEY WORDS: Abnormal frenal attachments, chondro-ectodermal dysplasia, Ellis–van Creveld syndrome, mulberry molars, polydactyly

Richard W.B. Ellis of Edinburgh and Simon van Creveld of Amsterdam first described Ellis–van Creveld (EVC) syndrome in 1940. EVC syndrome, also known as chondro-ectodermal dysplasia, is a rare autosomal recessive congenital disorder with an incidence of approximately 1 in every 60,000 live births and equal sex predisposition. A higher prevalence has been reported among certain populations like Old Order Amish and Arabs of the Gaza strip. Disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, a small chest, and a high frequency of congenital heart defects characterize this syndrome.[1] Oral manifestations include abnormal frenal attachments, congenitally missing primary and permanent tooth, microdontia, supernumerary tooth, and abnormal tooth morphology.[2]

Access this article online

Quick Response Code:

Website: www.jpbsonline.org

DOI: 10.4103/0975-7406.100257

Case Report

An 8-year-old boy reported with the chief complaint of missing upper and lower front teeth. This was his first dental visit and his medical history was non-contributory. He was the first child of non-consanguineous and normally developed parents. Pregnancy and birth were uneventful, though the newborn showed bilateral hexadactyly of his upper limb. He has a younger brother with no congenital abnormalities. The patient's stature likely appeared normal for his age; his head morphology, facial appearance, quantity and quality of the hair were also normal. The skeletal deformities like bilateral

How to cite this article: Jayaraj D, Maheswaran T, Suresh R, Ganapathy N. Ellis-van Creveld. J Pharm Bioall Sci 2012;4:153-6.

postaxial polydactyly, long and narrow appearing thorax compared to the height of lower limb, genu valgum, and short middle and distal phalanges were observed [Figure 1]. Other striking features included severely dystrophic finger and toe nails [Figures 2 and 3]. The patient's psychomotor and cognitive developments were within normal limits. Extraoral examination revealed slight V-notch on the middle part of the upper lip and mild defect in philtrum. The intraoral manifestations were remarkable. The labial frenum of the upper lip was hyperplastic and adhered to the anterior alveolar ridge obliterating the mucolabial sulcus [Figures 4 and 5]. Multiple lower labial freni attached to the anterior alveolar ridge were present with a serrated margin [Figure 6]. All primary and permanent incisors were clinically missing and there was no history of previous exfoliations/extractions of incisors. The erupted permanent canines were conical; primary and permanent molars of both arches were malformed with abnormal occlusal pattern resembling mulberry shape.

The patient was referred to a cardiologist to know the nature of cardiac involvement. The chest radiograph revealed a shadow of enlarged heart [Figure 7a]. An echocardiogram was done and a diagnosis for acyanotic congenital heart disease with large ostium primum atrial septal defect was confirmed. A hand-wrist radiograph revealed shortening of the middle and distal phalanges, and deformity in the fifth and sixth metatarsals and few carpal bones [Figure 7b]. Orthopantomogram revealed all permanent incisors were congenitally absent [Figure 7d]. The findings of routine blood investigations were within normal limits. Based on the overall clinical examination and the findings of various diagnostic investigations, a confirmatory diagnosis of EVC syndrome was established.

Treatment plan consisted of mucogingival surgery for excision of the labial frenulum in both upper and lower arch, followed by prosthetic rehabilitation for his missing teeth. Restorative care for carious teeth, professionally applied topical fluoride, and home care oral hygiene measures were also insisted with appropriate considerations for antibiotic prophylaxis for his underlying cardiac problem.

Discussion

EVC syndrome is a chondral and ectodermal dysplasia characterized by short ribs, polydactyly, growth retardation, and ectodermal and heart defects. This rare condition is inherited as an autosomal recessive trait with variable expression. Mutations of the EVC1 and EVC2 genes, located in a head-to-head configuration on chromosome 4p16, have been identified as causative. EVC belongs to the short rib-polydactyly (SRP) group, and these SRPs, especially type III (Verma-Naumoff syndrome), are discussed in the prenatal differential diagnosis. Postnatally, the essential differential diagnoses include Jeune dystrophy, McKusick–Kaufman syndrome, and Weyers' syndrome.[3] Recently, EVC has been included in a new class of human genetic disorders called "ciliopathies," where the underlying defect may be dysfunctional molecular mechanism in the primary cilia of cells.[4] Parental consanguinity has been confirmed in about 30% of cases.[4,5] Our patient was the first child of non-consanguineous and normally developed parents with no significant pre-natal, natal, and post-natal history, though the newborn showed bilateral hexadactyly of his upper limb. The patient had never sought any medical assistance previously regarding the same problem.



Figure 1: Long, narrow appearing thorax with knock knees



Figure 2: Bilateral hexadactyly of the upper limb with dytrophic nails



Figure 3: Dystrophic nails in the toes



Figure 4: Adherence of the upper labial frenum with the alveolar ridge obliterating the mucolabial sulcus and serrated margins of the lower alveolar ridge with multiple labial frenum



Figure 5: Missing incisors and malformed primary and permanent molars in the upper arch



Figure 6: Missing lower incisors and mulberry-shaped primary and permanent molars

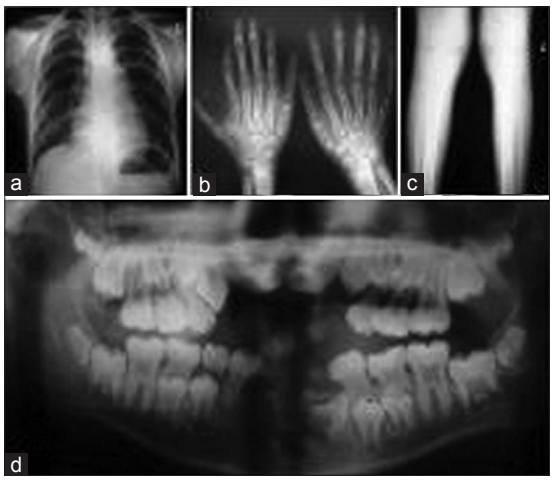


Figure 7: (a) Chest radiograph reveals shadow of enlarged heart. (b) Hand-wrist radiograph reveals shortening of the middle and distal phalanges. (c) Anteroposterior view of lower limb with genu valgum. (d) Orthopantomogram reveals congenital absence of all upper and lower incisors

EVC presents with characteristic tetrad: (1) disproportionate dwarfism with short limbs and exceptionally long trunk, (2) bilateral postaxial polydactyly of the hands, (3) dystrophic nails, hypodontia, and malformed teeth, and (4) congenital cardiac malformations occur in 50–60% of cases, the most common being the inter-septal defect.[6] Cardinal features after birth are short stature, short ribs, polydactyly, and dysplastic fingernails and teeth. The syndrome affects mainly the bones such as the lower limb with genu valgum, which requires surgical correction. The upper limbs show the characteristic bilateral postaxial polydactyly.[7] All these features were typically observed in our patient.

Cardiac defects, especially abnormalities of atrial septation, occur in about 60% of cases.[1,3,7-9] These patients die due to either cardiac problem or respiratory distress.[4] Our patient was diagnosed with large ostium primum atrial septal defect. Other ectodermal features in this syndrome are the absence or hypoplastic features of finger and toe nails, natal teeth, conical shaped, or missing teeth. These features were also observed in our case.

Bapaji[2] emphasized that oral manifestations in the EVC syndrome are characteristic and constant. The most constant finding is fusion of the anterior portion of the upper lip to the maxillary gingival margin, as a result of which no mucobuccal fold exists, causing the upper lip to present a slight V-shaped notch in the middle.[2,7,10] Changes in the upper lip are variously called partial hare-lip, lip tie, and so on. The anterior portion of the lower ridge is often serrated and presents with multiple small labial frenula. The maxillary and mandibular alveolar process presents with notching or submucous clefts and continuous or broad labial frenula and dystrophic philtrum. Bilateral incomplete clefts or notchings usually involve the region normally occupied by the lateral incisors, each submucous cleft being marked by a moderately sized fibrous band, whose fibers appear to incise the underlying alveolar process and extend across the mucobuccal fold into the lip.[2] The frenal attachments in our patient were typically as stated above. Veena *et al.*, stated that this could be due to the continuation

of the normal serrated condition of the gingiva from the third to seventh month in the uterine life of the fetus.[7]

Most frequent dental findings include neonatal teeth, partial anodontia, peg-shaped teeth, delayed eruption of teeth, and early involvement with caries.[10] Our patient had congenital absence of all primary and permanent incisors, which was confirmed by history and radiograph. Also, a remarkable feature observed in our patient was that all primary and permanent first molars were malformed with mulberry shape and the cuspids were conical shaped. Vinay *et al*. stated that the dental abnormality occurred in the initiation stage of tooth development and could result in the absence of teeth in the anterior region. A further insult at morphodifferentiation stage is probably responsible for the changes in size and form of the teeth. The changes in the anatomy of the crowns with deep fissures and pits could be a predisposing factor in the high rate of caries.[6]

Management of EVC is multidisciplinary. Management during the neonatal period is mostly symptomatic, involving treatment of the respiratory distress due to narrow chest and heart failure. Orthopedic follow-up is required to manage the bone deformities. Professional dental care should be considered for management of the oral manifestations. Prognosis is linked to the respiratory difficulties in the first months of life due to thoracic narrowness and possible heart defects. Prognosis of the final body height is difficult to predict. The management must be started at early age to achieve a good functional outcome.[1] Dental care in childhood includes dietary counseling, plaque control, and oral hygiene instructions. Crown or composite build-ups for microdonts can be fabricated. Partial dentures can be placed to maintain space and improve mastication, esthetics, and speech because of congenitally missing teeth. Malocclusion can be corrected with orthodontic treatment. Dental care during adulthood can be done with implants and prosthetic rehabilitation to replace congenitally missing teeth.[8]

Conclusion

An early diagnosis of EVC and therapeutic care with a multidisciplinary approach would warrant these patients in achieving a better quality of life care.

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Source of Support: Nil, **Conflict of Interest:** None declared.