CASE REPORT

Ellis–Van Creveld syndrome: Report of two cases

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Abstract

Ellis–van Creveld (EVC) syndrome is an ectodermal and chondral dysplasia as three embryonic layers are involved. It is a form of achondroplastic dwarfism resulting from an autosomal recessive disorder. It is most prevalent in the Amish population occurring in 1/5,000 live births and the birth prevalence in non-Amish population are estimated to be 7/1,000,000. Multiple malformations of the ectoderm and mesoderm, including short ribs, polydactyly, growth retardation and congenital cardiac anomalies are noted. This rare condition is inherited due to gene mutations of the EVC1 and EVC2, located on chromosome 4p16 in head to head configuration. The presence of multiple orodental findings such as conical teeth, hypodontia, malocclusion and hypoplasia of the enamel makes this syndrome important for a dentist. This article attempts to highlight on two female cases of EVC syndrome who reported to our institution where rare dental findings and skeletal features were diagnosed along with a review of the literature.

Keywords: Chondroectodermal dysplasia, hypodontia, multiple frenulas, postaxial polydactyly

Case Reports

Case 1

A 7-year-old girl child reported to our department with a chief complaint of missing teeth since childhood. She was the first born child of consanguineous parents who were apparently healthy, and her younger brother had no congenital abnormalities. Her past medical and surgical history revealed that she was a known cardiac patient diagnosed with atrial septal defect from birth and was under medications. She had undergone surgery for cleft lip at the age of one. The psychomotor and cognitive developments...
were normal. Though the child appeared to be short statured, [Figure 1a], her morphology of the head, facial appearance, quality and quantity of her hair and nails were normal.

General physical examination showed mild hypertelorism and strabismus with acromelic shortening of upper and lower limbs, bilateral postaxial polydactyly in fingers and toes, presence of hallux varus with knock knees.

Extra-oral examination showed a mild defect in the philtrum due to surgical scar. Intra-oral examination revealed hypodontia of primary and secondary dentition, multiple frenulas and notching in lower anterior alveolar ridge with absence of lower labial sulcus and conical shaped $1, 61$ and multiple carious teeth [Figure 2]. With the clinical findings the patient is subjected to radiographic evaluation.

Orthopantomography (OPG) radiograph reveals; all four permanent mandibular incisors and maxillary primary laterals including $75$ were congenitally absent and conical shaped upper primary incisors were seen. Transpositioned tooth bud of $35$ with $36$ erupted earlier in $75$ regions were also observed [Figure 3]. The hand wrist X-rays revealed postaxial (towards the $5^{th}$ finger side) polydactyly bilaterally and brachydactyly (short left $6^{th}$ and right $5^{th}$ fingers). Suspicious fusion of right $5^{th}$ and $6^{th}$ metacarpals, with no other obvious bony fusion, was seen. Mild inward curving of $5^{th}$ and $6^{th}$ finger with short middle phalanges in the respective right and left fingers suggestive of clinodactyly was observed [Figure 4a]. Foot radiograph findings confirmed, hallux varus (big toe is medially positioned resulting in widened space between the big toe and other toes) and left foot shows polydactyly [Figure 4b]. Lower limb radiograph shows knock knees, otherwise called as genu valgum (at the level of knee joint, they touch each other, but legs are pointing laterally) as shown in Figure 4c. Chest radiographs showed no evidence of cardiomegaly, while lung parenchyma is unremarkable/normal, with no evidence of short ribs.

Differential diagnosis includes McKusick–Kaufman syndrome, Weyer’s acrofacial dysostosis, Juene dystrophy ofofacial digital syndrome and EVC syndrome. The presented case was well correlated with the clinical and radiological features of EVC syndrome. The patient was advised for full mouth rehabilitation with antibiotic prophylaxis and has been referred to a multidisciplinary clinic for medical management.

Case 2

An 8-year-old girl reported to our department with a chief complaint of missing teeth in upper and lower front teeth region

Figure 3: Orthopantomography revealing the absence of permanent teeth buds of $31, 32, 41, 42$ and transpositioned teeth $35, 36$

Figure 4: (a) Radiograph of right and left hand wrist showing bilateral post axial polydactyly brachydactyly and clinodactyly; (b) foot radiograph reveals hallux varus and polydactyly of left foot, (c) radiograph of lower limb showing knock knees
since childhood. History revealed presence of natal teeth that was removed 2 days after her birth and since then teeth was missing in the same region. Her medical history revealed that she had cyanotic congenital heart disease, partial atrioventricular canal defect with cleft mitral valve and moderate mitral regurgitation, which was treated 2 years back. She was the first born child of parents with first degree consanguineous marriage, and her younger brother had a history of cleft lip.

General physical examination revealed that the child was conscious, co-operative, well-oriented with short stature, long thorax and short lower extremities [Figure 1b]. The skeletal deformities such as mild cubital valgus (deformity of the elbow in which it deviates away from the midline of the body when extended), postaxial bilateral polydactyly, syndactyly of 4th and 5th fingers and bifid distal aspect of 4th fingers were noted. Valgus deformity of the knee, acromesomelia (shortening of mesial and distal segment of limbs), wide space between hallux and other toes, plumpy lower extremities were present. Abnormal gait and dystrophic nails in all the fingers were noticed.

Extra-oral examination revealed strabismus (deviation of the eye from the normal axis), hypertelorism (increased width between eyes), and broad nose. Incompetent lips were present and middle portion of upper lip appeared to have the notch. Hard tissue examinations showed mixed dentition and missing teeth were upper and lower incisors along with multiple carious teeth. Soft tissue examination showed fusion of upper middle portion of the lip to the maxillary gingival margins, fusion of lower middle portion of the lip to the mandibular gingival margins and notching of lower alveolar process. Multiple frenulas were seen and absence of sulcus in the labial aspect of upper and lower anterior region were observed [Figure 5]. With the clinical findings the patient is subjected to radiographic evaluation.

OPG radiograph revealed missing upper and lower incisors and tooth bud of 15. Two unerupted supernumerary teeth were seen, one above 25 and the other between 45 and 46 [Figure 6]. The hand wrist X-rays revealed bilateral polydactyly, bifurcated middle phalanges of right 4th finger, fusion of capitate and hamate bones, shortened middle and distal phalanges of all fingers [Figure 7a]. The lower limb X-rays revealed hypoplasia of proximal lateral tibial epiphyses, irregular epiphyses and metaphyses with tilt deformities, valgus deformity (lateral deviation) of knees and defect in the lateral aspect of tibia [Figure 7b]. The chest X-rays revealed hypoplastic lateral ends of clavicle and presence of cardiomegaly [Figure 8].

Differential diagnosis includes Mckusick–Kaufman syndrome, Weyer’s acrofacial dysostosis, Juene dystrophy, orofacial digital syndrome and EVC syndrome. The presented case correlates positively to the features of EVC syndrome except for cleft lip or palate, mental retardation, and congenital
EVC is an autosomal recessive skeletal dysplasia which is characterized by short ribs, short limbs, postaxial polydactyly and dysplastic teeth and nails. Congenital heart defects happen in 60% of individuals. It does not show racial and gender predilection.[3,11,12] Prenatal abnormalities would be expected after the 18th week of gestation that includes narrow thorax, marked shortening of long bones, hands and feet hexadactyly and cardiac defect.[14]

The cardinal features after birth are increasing severity from the proximal to distal portions of the limbs with disproportionate small stature, shortening of middle and distal phalanges, hands affected with polydactyly, nails and teeth dysplasia, congenital heart malformations.[7] In both the cases reported here, all the tetrad of principal features was identified. Several additional findings reported include epi and hypospadias, strabismus, cryptorchidism and the thoracic wall malformation and pulmonary malformations and in rare cases renal abnormalities.[13] In both cases, strabismus was evident and in the second case, there were supernumerary teeth, hypoplastic clavicle where this uncommon finding appears unique and have not been much reported yet.

The essential radiological skeletal features observed in both cases were in accordance with previous reports which included fusion of the capitate and hamate bones of the wrist, knock-knees (defect of the lateral aspect of the proximal part of tibia) or genu valgum, cubitus valgus, hallux varus, clinodactyly of the fifth finger and fusion of 5th and 6th metacarpals. Other features present in addition to EVC syndrome were retarded bone formation, hypoplastic cubitus, supernumerary carpal bone center, and disturbances in bone remodeling of metacarpals/or phalanges.[9]

The most consistent oral findings in this syndrome is hypodontia involving maxillary and mandibular incisors which supports the aforesaid cases and also the visit of these patients made us to correlate with other features and proceed with radiological investigations to finally confirm this syndrome as EVC syndrome. Other oral findings in EVC syndrome are diverse, which not only affects the morphology of teeth but also soft tissues are labiogingival adherence, multiple frenulas, dystrophic philtrum, submucosal clefts, supernumerary teeth, abnormal or conical or microdontic teeth and malocclusion were also present in our case, which are the classical signs of EVC syndrome and made us to differentiate from other syndromes like Weyers acrofacial dysostosis, McKusick–Kauffman syndrome, Jeune dystrophy, orofacial digital syndrome.

Thorough search of the literature reveals that natal teeth have been reported only in 25-30% of cases,[14] as was seen in our second case. The first case presented here had transpositioned 36 and 35 and may be the first case to be reported and considered a rare entity. The clinical variability of oral EVC syndrome which is attributed to genetic and environmental phenotype modifying effects could be due to genetic factor on teeth and other oral structure development that occurs during a relatively long period.[9,12]

Confirmatory diagnosis is based on clinical manifestations of the syndrome, skeletal radiographic survey. DNA mapping is most reliable molecular diagnostic methods by direct sequencing of EVC and EVC2 genes,[13] however gene mapping was not attempted in our patients. Further studies are required to explicate other genes involved that may lead to various phenotypic manifestations of EVC syndrome.[15]

Though no definitive cure for EVC syndrome, effective management requires multidisciplinary approach such as clinical geneticist, cardiologist, orthopedician, physical and occupational therapist and since oral manifestation is one of the characteristic diagnostic features, the dentists play a major role in management of this syndrome related to esthetics and growth of the jaws.[16] Invasive dental procedures should be done under antibiotic prophylaxis. The removal of natal teeth should be done as they may impair the feeding. The patients and their parents should be given dental health education to emphasize on oral hygiene instructions, diet counseling, plaque control and frequent dental checkup, topical fluoride, and sealant application.

Conclusion

EVC syndrome poses a great challenge for the dentists in its identification, proper planning of the treatment and patiently treating them with more care. As a rare autosomal disorder, EVC syndrome can be diagnosed by its clinical and oral manifestations as presented. Apart from these constant clinical presentations our cases are reported with variable new features such as transposition of teeth, supernumerary teeth, hypoplastic clavicle, which can be considered as a new entity to this syndrome with their unique clinical significance.

References