ISSN: 0975-833X

Available online at http://www.journalcra.com

International Journal of Current Research
Vol. 8, Issue, 06, pp.33683-33685, June, 2016

INTERNATIONAL JOURNAL OF CURRENT RESEARCH

RESEARCH ARTICLE

ELLIS-VAN CREVELD SYNDROME – CASE REPORT WITH MULTIPLE DENTAL ANOMALIES

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ARTICLE INFO

Article History:
Received 24th March, 2016
Received in revised form
20th April, 2016
Accepted 15th May, 2016
Published online 30th June, 2016

Key words:

Chondroectodermal dysplasia,
Ellis-van Creveld syndrome,
Genu valgum, Polydactyly.

ABSTRACT

Ellis-van Creveld syndrome (EVC) also called as chondroectodermal dysplasia or mesoectodermal dysplasia is a rare autosomal recessive disorder caused by mutation in EVC1 and EVC2 genes (4p16). Classical EVC syndrome constitutes a tetrad of chondrodystrophy, ectodermal dysplasia, polydactyly and cardiac defects. Presence of variable clinical findings like fusion of upper lip to the gingival margin, presence of multiple frenula, conical, peg shaped and hypoplastic teeth, neonatal teeth, congenitally missing, absent or delayed eruption of teeth, molars with wide grooves and atypical cusps play an important role in the diagnosis of this syndrome. We report a case of 6 year old boy with short stature, genu valgum, bilateral postaxial polydactyly, dysplastic nails and multiple oral findings and dental anomalies. The diagnosis was made based on clinical and radiological features. EVC syndrome may be undiagnosed due to lack of awareness and improper screening. So, proper diagnosis and management is necessary.

Citation: Dr. Chaitanya, P., Dr. Sharada Reddy, J., Dr. Suhasini, K. and Dr. Hema Chandrika, I. 2016. "Ellis-van Creveld Syndrome – Case Report with Multiple Dental Anomalies", *International Journal of Current Research*, 8, (06), 33683-33685.

INTRODUCTION

Ellis-van Creveld (EVC) syndrome also called as chondro-ectodermal dysplasia or meso-ectodermal dysplasia was first described by Richard WB Ellis and Simon van Creveld of Amsterdam in 1940 (Ellis and Van Creveld, 1940). It is a rare autosomal recessive disorder resulting from a genetic defect located on chromosome 4p16. Two different mutations EVC1 and EVC2 have been identified (Tompson *et al*., 2007; Arya *et al*., 2001). In 30% of the cases parental consanguinity have been reported (Cahuana *et al*., 2004). Classical EVC syndrome presents a tetrad of chondrodystrophy, polydactyly, ectodermal dysplasia and cardiac defects. However, variable clinical manifestations could be frequently present. We report a case of 6 year old boy with multiple oral findings and dental anomalies.

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Case Report

A 6 year old boy, product of first degree consanguineous marriage, born of full-term normal delivery, presented with complaint of deformity of both the legs and short stature. Antenatal, natal and post natal histories were non contributory except for the presence of bilateral polydactyly of hands since birth. On general examination, child was conscious, coherent, cooperative with normal intelligence. Stature was 101.6 cm which was relatively short for his age with disproportionate short limbs compared to axial skeleton. Length of upper segment of the body was more compared to lower segment. Narrow thorax was present. Upper limbs were short and plump with sausage shaped fingers. Middle phalanges of hands were short and broad. Bilateral manual postaxial polydactyly of hands was seen with supernumerary finger on ulnar side.

Lower limbs were deformed with outward bending of knees (bilateral genu valgum). Nails were thin, hypoplastic and irregularly shaped (Figure 1). Scalp, eyebrows were normal. Hair was fine and straight but not sparse. No cardiac, genitourinary, hepatic or CNS abnormality was seen on clinical examination. Intraoral examination revealed fusion of mid portion of upper lip to maxillary gingival mucosal margin leading to obliteration of vestibule and absence of mucobuccal fold. Thick and broad labial frenum extending from lower lip was seen producing a notch on mandibular ridge. Notching was also seen on the maxillary anterior segment. Multiple small upper and lower labial frenula were present (Figure 2). In the maxillary arch conical right lateral incisor (52), rotation of right central incisor (51), fusion of left central (61) and lateral incisors (62) was found with presence of talons cusp on palatal aspect of 51 and 62. Atypical molars were seen with wide grooves and atypical cusps. In the mandibular arch, anterior hypodontia was seen with congenitally missing primary central incisors (71, 81) along with microdontic and hypolastic lateral incisors (72, 82) and a supernumerary tooth between left lateral incisor (72) and canine (73). Deep dental caries was seen in molars (Figure 3).

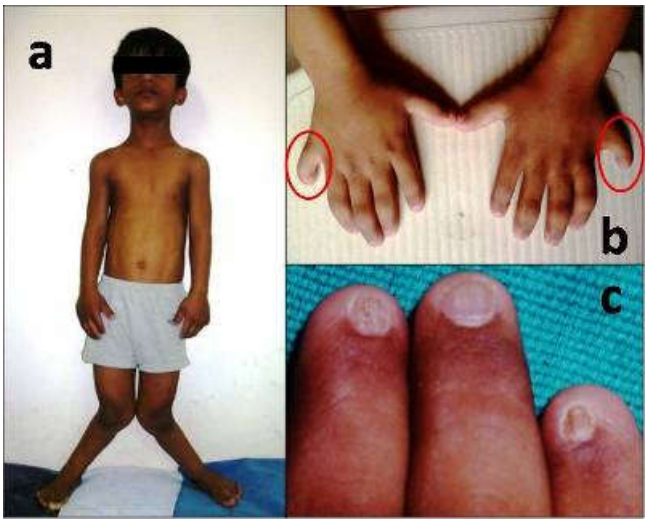

Figure 1. a) Short stature, genuvalgum b) hexadactyly and c) hypoplastic nails of hands

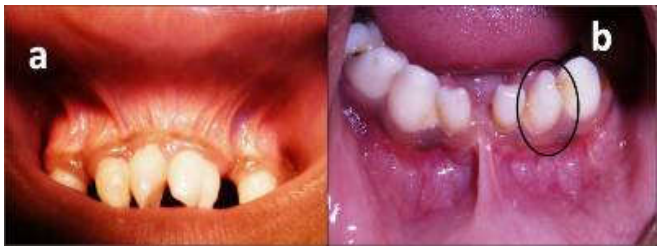

Figure 2. a) Multiple labial frenula, conical right lateral, fusion of left central and lateral incisors. Note alveolar notches on ridge. b) Thick labial frenum, missing lower central incisors and a supplemental tooth between left lateral incisor and canine

Investigations

Vital data was normal. There was no abnormality on evaluation of gastrointestinal, cardiovascular, respiratory and musculoskeletal systems. No abnormality in kidneys were detected. Radiographs of upper limbs revealed curved humerus, polydactyly of both hands, fused metacarpels of 5th & 6th digits. Middle phalanges were broad and short with cone shaped epiphysis. Radiographs of lower limbs revealed acromesomelia (relative shortening of middle and distal segment of limbs), hypoplasia of upper lateral tibial epiphysis with knock knee.

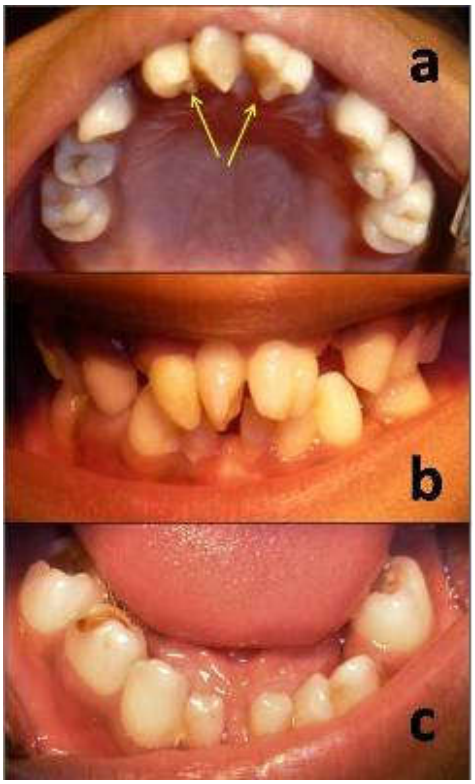

Figure 3. a) Maxillary occlusal view showing molars with wide grooves and atypical cusp. Arrows indicate talons cusp. b) Anterior teeth malocclusion c) Note deep caries in lower molars

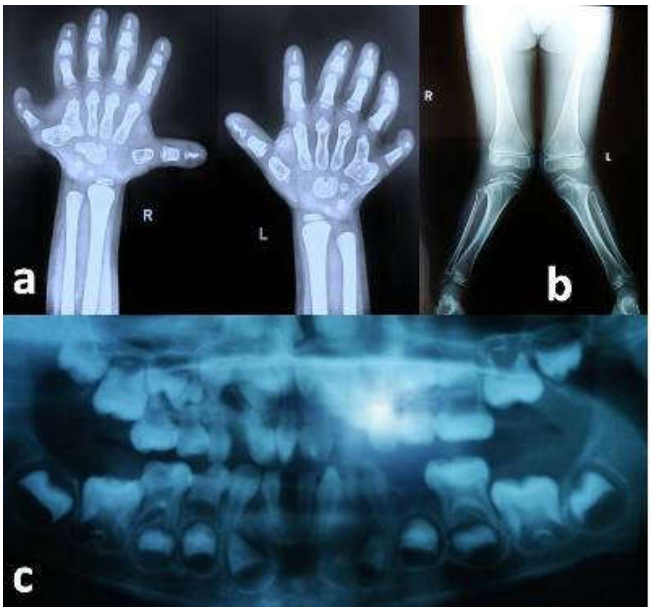

Figure 4. Radiographs of a) hand showing polydactyly, fused metacarpals of 5th and 6th digits b) legs showing knock knees, hypoplasia of upper lateral tibial epiphysis c) panoramic radiograph showing congenitally missing permanent incisor tooth buds, taurodontism in molars

Shortening is pronounced more in tibia and fibula than femur. Fibula is disproportionately shorter than the tibia. Panoramic radiograph revealed absence of permanent lower central (31, 41) and lateral incisor (32,42) tooth buds. Taurodontism was present in primary molars (Figure 4). Association of short stature, polydactyly of hands, oligodontia, dysplastic nails along with other clinical and radiologic features lead to the diagnosis of EVC syndrome.

DISCUSSION

EVC syndrome is a tetrad with chondrodysplasia, polydactyly, ectodermal involvement and congenital heart defects. Chondrodystrophy is disproportionate dwarfism. Patients also have lumbar lordosis and genu valgum. Polydactyly is usually bilateral and on ulnar side. In upto 93% of cases ectodermal dysplasia involving nails and teeth was observed. Hypoplastic, dystrophic, friable or even absent nails may be present. Tooth involvement may include neonatal teeth. Congenitally missing primary and permanent teeth, microdontic, conical or peg shaped, hypoplastic teeth, molars with abnormal cusps or accessory grooves, teeth with talons cusp, supernumerary teeth, fusion of teeth and taurodontism (enlarged pulp chamber) may be seen as observed in this case along with other findings like absent or delayed eruption, premature loss of teeth, malocclusions (Shekhar *et al*., 2011; Joshi *et al*., 2015), serrated incisal margins, dental transposition (Gopal and Belavadi, 2014), dysmorphic conical shaped roots (Cahuana *et al*., 2004). Other common oral manifestations include fused mid portion of upper lip to maxillary gingival margin so that no mucobuccal fold exists, causing upper lip to present a slight V-notch in middle (Atasu and Biren, 2000). The anterior portion of mandibular alveolar ridge is often serrated, and multiple small labial frenula are frequent (Cahuana *et al*., 2004; Atasu and Biren, 2000). All of these features were observed in this case. Hair is usually normal, but occasionally sparse, thin, brittle, hypochromic (Harper and Trembath, 2004). Common congenital heart malformations include atrial septal defect, single common atrium, patent ductus arteriosus, hypoplasia of aorta and rarely ventricular septal defect (Joshi *et al*., 2015). Other anomalies include musculoskeletal anomalies like low set shoulders, narrow thorax, broad hands & feet and sausage shaped fingers. Occasionally there are genitourinary abnormalities. Liver and CNS abnormalities may also be present (Harper and Trembath, 2004).

The diagnosis is essentially clinical based on manifestations described and supported by skeletal and radiological survey. Antenatal diagnosis can be made during 13th week of gestation when the increased nuchal translucency is evident. Other skeletal features like narrow thorax, cardiac defects, marked shortening of long bones, hexadactyly of hands and feet can be seen on ultrasound images done around 18th week of gestation (Gopal and Belavadi, 2014). The longevity of survival depends on severity of the underlying heart disease. Most of the deaths are directly related to the cardio-respiratory compromise (Gopal and Belavadi, 2014). As there was no underlying cardiac abnormality, normal life span could be expected in this child.

Recurrences may be prevented by genetic counseling, amniocentesis and/or chorionic villus sampling to demonstrate the mutations in EVC genes in the first trimester of pregnancy. A multidisciplinary approach is advised involving a clinical geneticist, cardiologist, pulmonologist, orthopedician, physical and occupational therapist, dentist, pediatrician and pediatric neurologist for proper management and rehabilitation of these cases and reduce to their morbidity (Shekhar and Kumud, 2011). Orthopedic follow up is required to manage bone deformities. Early treatment helps to reduce psychological trauma to the patient.

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