ELLIS VAN CREVELD SYNDROME- A CASE REPORT

ABSTRACT: Ellis Van Creveld Syndrome (EVC) is a infrequent inborn genetic disorder with an x-linked recessive pattern of inheritance. This is recognized by bilateral accessory little finger in upper extremity, short limbs, ectodermal dysplasia affecting teeth and nails, and congenital cardiac defect. The overall prevalence of this disease is 7 in 1000000. In this current case, a 9year old female patient reported with typical general and oral manifestations, which were key diagnostic features of EVC syndrome.

KEYWORDS: Ellis van Creveld syndrome, Congenital heart defect, autosomal recessive

INTRODUCTION: EVC is a mesenchymal ectodermal dysplasia first reported by two Pediatricians named Richard W.B Ellis and Simon van Creveld in 1940[1]. It is a infrequent congenital x-linked recessive syndrome due to a genetic abnormality on chromosome 4p16[2]. It is brought by a mutation in EVC1 and EVC2 genes with parental consanguinity in 30% of cases[3,4,5]. In 1964, Mukusick et al reported many affected individuals in the Amish community of Lancaster Country, Pennsylvania of USA and in western part of Australia. The frequency of this disorder in the Amish population is 1 in 5000 actual births, whereas in the other communities is 7 in 1000000[4,5].

EVC Syndrome presents with the following tetrad of clinical manifestations[6] which includes; First most typical hallmark triat is skeletal dysplasia of the os longum, which results in a relatively small stature and elongated torso,resulting in a hazardous ossification defect[5]. The intensity of short limbs rises as they progress from proximal to distal. Lateral ray polydactyl of upper extremities with a extra digit usually begins on the side of little finger. With large hands and feet,sausage-shaped fingers are common. Cloutson syndrome with dystrophy of nails, weak and scanty hair, and oral manifestation[7]. In 50-60% of cases, there is inborn heart illness, the most prevalent of which involves only one atrium chamber and ventricular septal defect[5].

CASE REPORT: A 9-year-old young female patient who reported to the Department of Oral Medicine and Radiology with a one-year history of disparity among her upper and lower front teeth. Exfoliation of primary dentition and lack of permanent teeth eruption in the upper anterior region was also mentioned by the patient. Medical history revealed congenital heart disease (Arterio ventricular canal defect) for which she had undergone cardiac surgery at the age of 1 year. There is no history of the parents marrying consanguineously and family history is also noncontributory.

The child had a small stature and both limbs are shortened and malformed on general inspection. (Fig.1). She also presented with strabismus (Fig.2), cubitus valgus, narrow chest, hypoplastic fingernails, the big toe and the other toes have a significant disparity between them in the right foot (Fig.4). The patient cognitive capability was within a reasonable range. The intraoral evaluation divulged morphological alterations in upper and lower anterior, midline diastema, highly attached labial frenum in the maxillary anterior region, absence of both maxillary lateral incisors (Fig.3).

Panoramic radiograph confirmed the absence of maxillary laterals (Fig.5.A). Shortening of the extremities, genu valgum, has been seen on a radiograph of the upper and lower limbs(knock knees), bilateral postaxial polydactyly, hand wrist radiograph showed the hamate and capitate bones, as well as

the 5th and 6th metacarpal bones, are fused together (Fig.5.B). On the basis of clinical and radiological evidence with an associated congenital heart defect, the patient was provisionally diagnosed with Ellis van Creveld syndrome.

DISCUSSION: Chondroectodermal dysplasia is a infrequent x-linked recessive disorder. When enhanced nuchal translucency is observed on ultrasound, the detection of this disease can be confirmed as soon as the 18th week of pregnancy [4]. The prevailing clinical sign is acromesomelic dwarfism owing to defect in ossification affecting tubular bones, which emanates in the shortening of bones of limbs[8]. Other attributes consists of polydactyl usually bilateral accessory little finger which is commonly present in upper limbs and on the ulnar side of limbs and only in 10% of reports, it can be present in lower limbs[3]. EVC individuals also had cubitus valgus, genu valgum, dysplastic fingers and nails, and narrow chest [4]. Inborn cardiac defects are present among 60% of cases which include defects of bicuspid and right atrioventricular valves, ductus arteriosus closure, atrial and ventricular septal defect which are leading reasons for reduced life expectancy[9]. Some inconstant clinical features of EVC are congenital cataracts, nephrocalcinosis, strabismus, retinitis pigmentosa, central nervous anomalies, hypoplastic penis, and hematological anamolies[10].

The characteristic oral findings of this syndrome which aids in diagnosis which are, union of upper lip to the gingival margin, leading to loss of mucobuccal fold, partial non union of upper lip, multiple small accessory frenum, tongue tie, malignment of teeth, atypically small teeth, missing of lower successor anterior teeth and enamel hypoplasia[11]. In our present case, most of the clinical signs and symptoms such as acromesomelic dwarfism, bilateral postaxial polydactyl, narrow chest, cubitus valgus, hypoplastic fingers and nails, genu valgum, and previously treated congenital heart disease were seen. In addition to this, some inconstant features such as strabismus and unilateral postaxial polydactyl in the left foot and anodontia of maxillary laterals are not reported in much of the literature. The definitive diagnosis of this syndrome is genetic-based where homozygosity for mutation in EVC1and EVC2 genes which is detected by direct sequencing. However, the genetic mutation is positive only in 2/3rd of patients[12]. Due to less availability of genetic studies, the diagnosis of the current case was arrived by clinical and radiological findings. Although inborn cardiac defect and breathing illnesses are the leading causes of death in this disorder, those who sustain oneself childhood will get a standard survival rate. The individual who survived the oldest was eighty two years old[13].

Differential diagnosis of this case was curry hill syndrome, Jeune syndrome, orofacial digital syndrome, Morquio syndrome, and achondroplasia. Since there is no cure for EVC[14], Management focuses on symptomatic relief followed by a multidisciplinary approach. Some studies have found somatotropin insufficiency in this disorder, along with supplementation of somatotropin has been shown to improve these patients' stature[15].

CONCLUSION: EVC Syndrome is a occasional congenital genetic disorder which needs a multidisciplinary approach for a suitable diagnosis and management. Patients with this syndrome encounter a high mortality index because of cardiovascular and respiratory complications. Hence dentists play an essential role in early detection and establishing management protocols. Dentists can help

improve the esthetics and function, overcoming psychological trauma, and enhance the wellbeing of patient.

PATIENT CONSENT: The authors attest to having gotten all necessary patient consent papers. The patient(s) mother has/have consented in the form for her daughter's photos and other diagnostic evidence to be published in the paper. The consent individual mother is aware that their identities and surnames will be not be advertised, and that while every effort will be taken to keep their identities hidden, privacy could not be guaranteed.

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FIGURE.1. Patient with short stature measuring about 119cm in height

FIG.2. Patient show strabismus, on clinical examination.

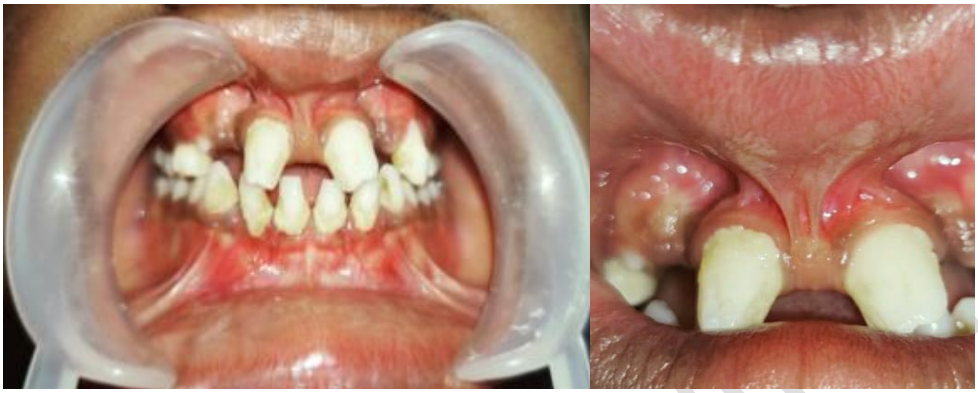

FIGURE.3. On intraoral examination, conically shaped tooth in upper and lower anterior. The frenal fibers stretch upto the palatine papilla,crossing the alveolar process.

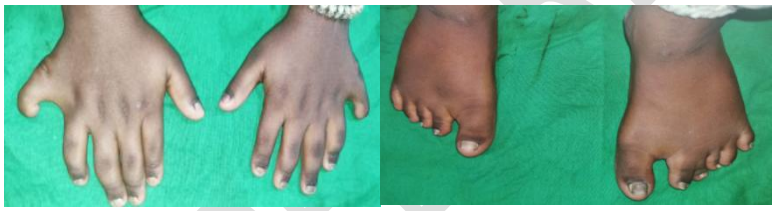

FIGURE.4. Extra digit is evident on both upper and lower limbs with hypoplastic nails and the big toe is separated from the other digits by a significant distance.

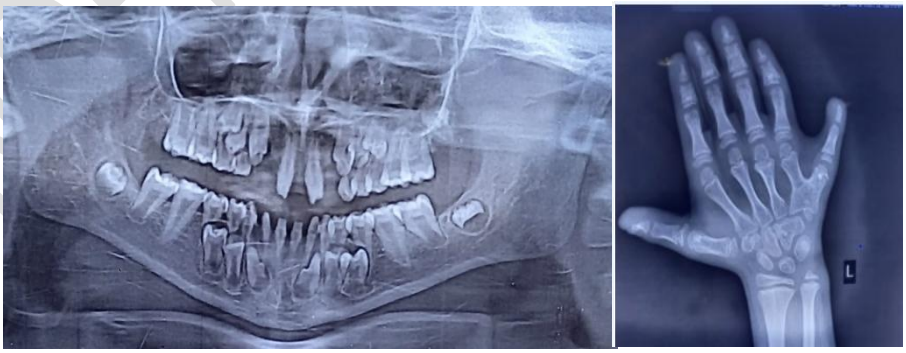

FIGURE.5.A.AND.B. Orthopantogram confirms conical-shaped maxillary and lower front teeth with congenitally missing maxillary laterals. The hand-wrist X- ray depicts union of carpals and extra digit on side of little finger.