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Ellis Van Creveld Syndrome: A Rare Case Report

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Abstract
Ellis Van Creveld syndrome (EVC syndrome) is a rare autosomal recessive disorder characterized by short stature, postaxial polydactyly, ectodermal dysplasia, and congenital heart defects. A 10-year-old female patient reported with disparity between the upper and lower anterior teeth, delayed exfoliation of primary teeth, and delayed eruption of permanent anterior teeth. Medical history revealed an atrial septal defect with exertional breathlessness. General examination showed short stature, hypoplastic nails, bilateral postaxial hexadactyly, and genu valgum. Intraoral examination revealed multiple missing teeth, morphological alterations of mandibular anterior teeth, and multiple frenula. Radiographic examination demonstrated congenitally missing mandibular incisors and canines, impacted maxillary anterior teeth, taurodontism, shortening of extremities, fused carpal bones, and genu valgum. Based on clinical and radiographic findings, a provisional diagnosis of Ellis–van Creveld syndrome was made. This case highlights the importance of oral and radiographic features in early diagnosis of this rare condition.

Keywords: *Ellis Van Creveld syndrome (EVC syndrome); EVC syndrome; Hypodontia; Taurodontism; Polydactyly; Congenital heart disease; Radiographic features.*

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I. Introduction

Ellis van creveld syndrome (EVC syndrome), also known as chondroectodermal dysplasia, is a rare autosomal recessive disorder characterized by a distinctive tetrad of abnormalities involving the skeletal system, ectodermal structures, and cardiovascular system. The syndrome was first described in 1940 by Richard W.B. Ellis and Simon van Creveld, who identified the association of ectodermal dysplasia, polydactyly, chondrodysplasia, and congenital heart disease as a unique clinical entity[1]. EVC syndrome primarily affects derivatives of multiple embryonic germ layers and is now recognized as a multisystem developmental disorder. The core clinical features include: Disproportionate short limbed dwarfism (acromelic type) due to defective endochondral ossification, Postaxial polydactyly, most commonly affecting the hands, Ectodermal dysplasia, manifesting as nail hypoplasia, abnormal dentition, and sparse hair, Congenital heart defects, particularly atrial septal defects or common atrium[2]. The syndrome follows an autosomal recessive inheritance pattern, as established through genetic studies and familial clustering, particularly among consanguineous populations[3]. A high prevalence has been reported among the Amish community, further supporting its recessive mode of inheritance[2]. Oral and dental manifestations are among the most diagnostically significant features of syndrome. These include: Multiple frenula, Obliteration of the labio-gingival sulcus, Hypoplastic and conical teeth, Partial anodontia, Enamel hypoplasia. Such oral findings are often considered pathognomonic and assist in differentiating EVC syndrome from similar syndromes such as orofacial digital syndrome and asphyxiating thoracic dystrophy[2]. Cardiac involvement is seen in approximately 50–60% of cases, commonly presenting as a common atrium or atrioventricular canal defect[4]. These cardiac anomalies significantly influence prognosis and early mortality. Beyond the classical tetrad, additional systemic involvement has been reported. Histopathological studies demonstrate renal medullary dysplasia, bile duct dysplasia, and abnormalities in growth plate cartilage, indicating that the syndrome affects derivatives of all three germ layers[5]. Similarly, overlapping phenotypes with Jeune syndrome and renal hepatic pancreatic dysplasia suggest that these entities may represent a disease spectrum rather than completely distinct disorders[6]. Prenatal diagnosis has also been reported using fetoscopy and ultrasonography, particularly in families at risk, highlighting the importance of genetic counselling and early detection[3].

II. Case Report

A 10 year old female patient reported who reported to the department of oral medicine & Radiology with complaint of one & half year disparity among her upper & lower front teeth. Delayed exfoliation of primary dentition of the lower anterior region & delayed eruption of upper anterior region. After detailed medical history revealed congenital heart disease (Arterial septal defect) for which arterial septal defect closure was planned. Patient also complains of difficulty in breathing while climbing stairs. There was no history of the parents marrying consanguineously & family history is also non-contributory.On general inspection the child had small stature thick both upper & lower limbs, broad skull (Fig.1). She also presented with hypoplastic upper lip, bilateral post axial hexadactyly, hypoplastic finger nails (Fig.2), knock knees. Patient cognitive capability was within a reasonable range. The intraoral examination revealed congenitally missing lateral incisors in mandibular anterior, clinically missing maxillary both lateral incisors & left central incisor Morphological alteration of lower anterior teeth, multiple frenum in maxillary anterior region with high frenal attachment (Fig.3). Panoramic radiograph confirmed the absence of mandibular permanent central incisors, both lateral incisors & both permanent canines. She also had impacted maxillary both central incisors & left lateral incisor with taurodontism of maxillary & mandibular first permanent molars bilaterally and maxillary second primary molars bilaterally (Fig.4). Hand wrist radiograph showed shortening of extremities, bilateral postaxial polydactyly, bilaterally fussed 5^{th} & 6^{th} metacarpal with bilaterally fussed hamate capitate bone (Fig.5). Lower libs radiograph showed genu valgum (Fig.6). On the basis of clinical radiological evidence with an associated congenital heart defect, patient was provisionally diagnosed with EVC syndrome.

III. Discussion

Ellis–van Creveld syndrome (EVC syndrome), also known as chondroectodermal dysplasia, is a rare autosomal recessive disorder first described by Ellis and van Creveld in 1940. The syndrome is characterized by a classical tetrad consisting of disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, and congenital cardiac anomalies. The estimated prevalence of Ellis–van Creveld syndrome is approximately 7 per million live births, although higher frequencies have been reported in isolated populations[7].

Ellis van Creveld syndrome is caused by mutations in the EVC 1 and EVC 2 genes located on chromosome 4p16, and parental consanguinity has been reported in approximately 30% of cases[2]. Clinically, the syndrome primarily affects structures derived from mesoderm and ectoderm, producing characteristic skeletal abnormalities such as short limb dwarfism, narrow thorax, genu valgum, and postaxial polydactyly of the hands and feet. These skeletal abnormalities are considered important diagnostic criteria[2]. Congenital cardiac defects are reported in approximately 50–60% of affected individuals, most commonly atrial septal defect or common atrium, and represent the major cause of morbidity and mortality in these patients[2,4]. Oral manifestations are highly characteristic and play a significant role in early diagnosis. The most frequently reported findings include: Hypodontia or partial anodontia, Delayed eruption of teeth, Conical or peg shaped teeth, Enamel hypoplasia, Taurodontism, Serrated alveolar ridges, Multiple accessory labial frenula, Absence of mucobuccal fold, Malocclusion. These findings have been consistently documented in the literature and are considered important diagnostic indicators of the syndrome[2,8]. Among these manifestations, absence of mucobuccal fold and multiple frenula are considered characteristic features and may result from abnormal development of the alveolar processes and vestibular sulcus during embryogenesis[2]. Radiographic examination typically reveals congenitally missing teeth, delayed tooth bud formation, taurodontism, and abnormal root morphology, which assist in confirming the diagnosis[2]. The differential diagnosis of EVC syndrome includes several syndromes with overlapping skeletal and dental features such as Weyers acrofacial dysostosis, Jeune syndrome, and McKusick Kaufman syndrome. However, the presence of the classical tetrad helps in establishing the diagnosis.

Early diagnosis is essential because dental professionals may be the first to identify the oral manifestations of the syndrome. Recognition of these features allows early referral for systemic evaluation and appropriate management[2,4].

Our present case showed several classical systemic features including short stature, thickened limbs, broad skull, hypoplastic upper lip, bilateral postaxial hexadactyly, hypoplastic fingernails, and genu valgum, which are characteristic skeletal and ectodermal manifestations of Ellis–van Creveld syndrome. The cognitive development of the patient was normal, which is consistent with previous reports indicating that intelligence is usually unaffected in this syndrome. Congenital heart disease is one of the most important systemic manifestations of EVC syndrome and occurs in nearly half of affected individuals. The presence of atrial septal defect and associated breathing difficulty on exertion in the present patient further supports the diagnosis and indicates systemic involvement. Dental and oral manifestations play an important role in the diagnosis of EVC syndrome. The patient presented with delayed exfoliation of primary teeth and delayed eruption of permanent teeth, which are commonly reported findings in this syndrome. The presence of multiple missing teeth, particularly mandibular incisors and maxillary lateral incisors, is considered one of the most characteristic dental findings and has been frequently reported in previous case studies. Morphological alterations of mandibular anterior teeth observed in

this patient are consistent with ectodermal dysplasia–related dental abnormalities described in EVC syndrome. The presence of multiple frenula with high frenal attachment in the maxillary anterior region represents another characteristic oral feature and is believed to result from abnormal development of the alveolar processes and vestibular sulcus. Radiographically, the patient showed multiple impacted teeth and extensive hypodontia, which are commonly reported findings in EVC syndrome. The panoramic radiograph demonstrated congenital absence of mandibular permanent incisors and canines along with impacted maxillary anterior teeth, indicating severe disturbance in tooth development.

Another significant radiographic finding in this patient was taurodontism involving both maxillary and mandibular first permanent molars and maxillary second primary molars, which has been described as a frequent dental anomaly in EVC syndrome and reflects abnormal root development.

Hand-wrist radiographs revealed shortening of extremities, bilateral postaxial polydactyly, fusion of the fifth and sixth metacarpals, and fusion of hamate and capitate bones, which represent typical skeletal abnormalities associated with this syndrome. Lower limb radiographs confirmed genu valgum deformity, another commonly reported skeletal feature. Due less availability of genetic studies and financial constraints the diagnosis was arrived mainly on clinical and radiological findings. Congenital heart defects and respiratory problems are the primary causes of mortality in EVC syndrome; however, patients who survive childhood usually have a normal life expectancy. The oldest reported patient survived up to 82 years of age[2,3].

Since there is no cure for EVC syndrome management focuses on symptomatic relief followed by multidisciplinary approach involving paediatricians, cardiologists, orthopaedic surgeons, and dental specialists. Dental management focuses on preventive care, restoration of carious teeth, prosthetic rehabilitation, and orthodontic correction to improve function and aesthetics[2,9].

Abbrevations: EVC syndrome - Ellis van Creveld Syndrome, Fig – Figure,

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Figures:

Fig.1 Showing short stature brachycephalic skull with knock knees.

Fig. 2 Showing bilateral post axial hexadactyly, hypoplastic finger nails

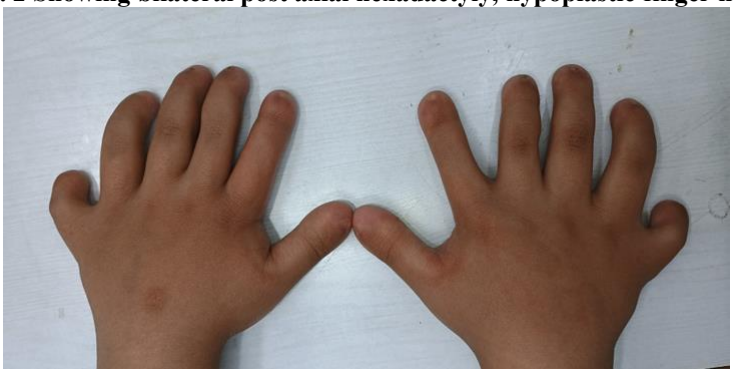

Fig.3 Showing multiple frenum in maxillary anterior region

Fig.4 Showing congenitally missing mandibular permanent incisors with taurodontism of permanent first molars (yellow arrow) of maxillary and mandibular jaw with primary maxillary second molars(red arrow).

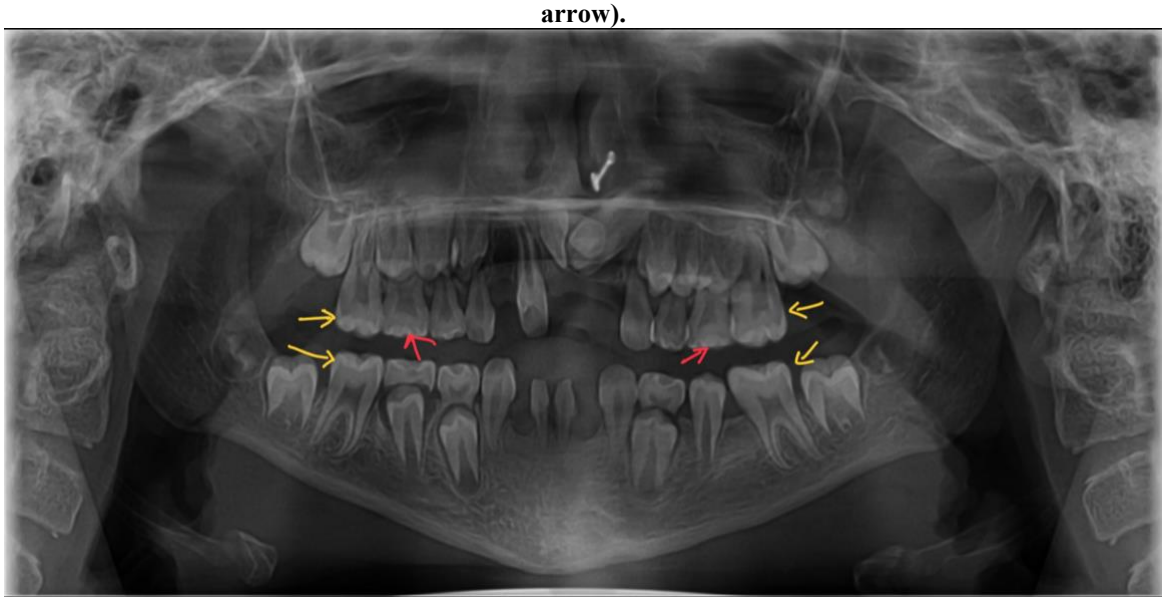

Fig.5 Bilateral postaxial polydactyly, bilaterally fussed 5th & 6th metacarpal with bilaterally fussed hamate capitate bone

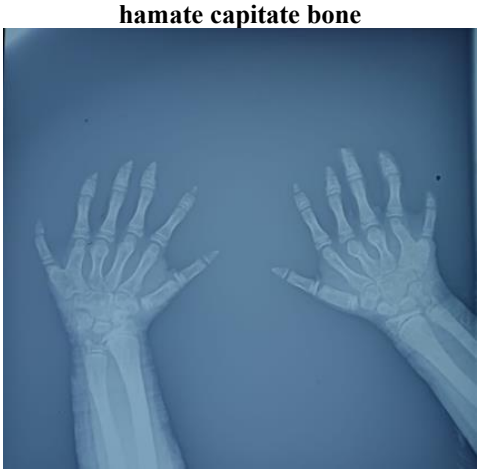

Fig.6 Disproportionately short femur and tibia and genu valgum