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Case Report

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# MANAGEMENT OF ORAL MANIFESTATIONS OF ELLIS VAN CREVELD SYNDROME: A CASE REPORT

**[1]Prof. Divya S. Sharma, [2*]Dr. Ashik Mohamed VN and [3]Dr. Pranshu Varshney**

[1]Professor, Department of Pediatric and Preventive Dentistry, Dr Ziauddin Ahmad Dental College, A.M.U., Aligarh.
[2,3]Resident Doctor, Department of Pediatric and Preventive Dentistry, Dr Ziauddin Ahmad Dental College, A.M.U., Aligarh.

***Corresponding Author: Dr. Ashik Mohamed VN**

Resident Doctor, Department of Pediatric and Preventive Dentistry, Dr Ziauddin Ahmad Dental College, A.M.U., Aligarh.

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## ABSTRACT

A rare autosomal-recessive disorder called Ellis-van Creveld syndrome is characterized by short limbs, post-axial polydactyly, ectodermal dysplasia, an edentulous mandibular incisor region, the absence of a mucobuccal fold, congenitally missing teeth, light alveolar ridge serrations, and numerous small alveolar notches. This clinical report includes findings such as polydactyly, clinodactyly, hypoplastic nails, congenitally missing multiple teeth, serrations in the incisors and alveolar notches. The case report also emphasizes on patients' preventive and therapeutic oral health care as well as differential diagnosis. Management of Ellis-van Creveld syndrome involves dentist, nutritionist, radiologist, pediatrician, cardiologist, orthopaedician, psychologist, geneticist for the positive outcome of the disorder. Recognizing the oral findings is important for early diagnosis and correct treatment planning of the syndrome. Various treatment strategies include oral prophylaxis, preventive measures for dental caries, restoration of dental caries, prosthodontic rehabilitation, orthodontic correction of malocclusion, and surgical corrections of soft tissue and skeletal abnormalities which must be followed up regularly by a pedodontist.

**KEYWORDS:** Ellis-van Creveld syndrome, Chondroectodermal dysplasia, Dental anomalies.

## INTRODUCTION

Chondroectodermal dysplasia, commonly known as Ellis-van Creveld syndrome, was initially described by Richard W.B. Ellis and Simon van Creveld in 1940.[1] The syndrome is because of mutation in two genes EVC1 and EVC2, both of them mapping on chromosome 4p16, in a head to head configuration.[2] The Old Amish Community used to experience this disorder frequently, but it is now found to affect people of all races. The estimated birth prevalence in the general population is 7/1,000,000. 30% of the cases were reported to have parental consanguinity.[3]

Ellis-van Creveld syndrome presents with a characteristic tetrad of clinical manifestations

i. Chondrodysplasia of the long, tubular bones which causes disproportionate dwarfism, and the severity of short limbs increases from the proximal to the distal portions.
ii. Bilateral polydactyly of the hands. Supernumerary finger is usually located on the ulnar side. Fingers appear to be sausage shaped, and the hands and feet are found to be wide.[4]
iii. Features of Hidrotic ectodermal dysplasia which includes dystrophic, small dysplastic nails, thin sparse hair, and oral manifestations.
iv. Congenital cardiac abnormalities can occur in 50% to 60% of cases, with single atrium and ventricular septal defect being the most prevalent defect. The related cardiorespiratory disorders are thought to be the primary factor in these individuals' shorter lifespans.[5]

These patients have a variety of oral manifestations, including hyperplastic frenula, mucobuccal fold absence, alveolar ridge serrations, multiple small alveolar notches, partial cleft lip, neonatal teeth, partial anodontia, conical and microdontic teeth, enamel hypoplasia, delayed teeth eruption, and taurodontism. Malocclusion can be present which are secondary to oral abnormalities and it is of no specific type.[6]

### Case report

An eight year old female patient reported to the department with the chief complaint of delayed eruption of lower front teeth. History of the present complaint revealed that non eruption of teeth was noticed around 1 year. History also revealed no primary teeth was erupted in lower front teeth region. Family history revealed that the girl was the child of consanguineously married and normally developed parents. Prenatal history was

uneventful with full term gestation period and normal delivery.

Upon general examination, patient had short statured but normal built and gait, with a height of 105 cm, weight of 18 kg and head circumference of 45.5cm (fig 1). The patient also had polydactyly of both hands, sausage-shaped digits, clinodactyly (fig 3). Lumbar lordosis was also noticed (fig 1a). Nails were found to be hypoplastic (fig 3c). Hair was found to be thin and friable and scalp showed traces of fungal infection (fig 3d). Facial morphology was found to be normal with straight profile and bilaterally symmetrical. Lips were competent (fig 2). Temporomandibular joint movements were found to be normal and tenderness and clicking sounds were absent.


**Fig. 1: Showing Frontal and Lateral view of the patient.**

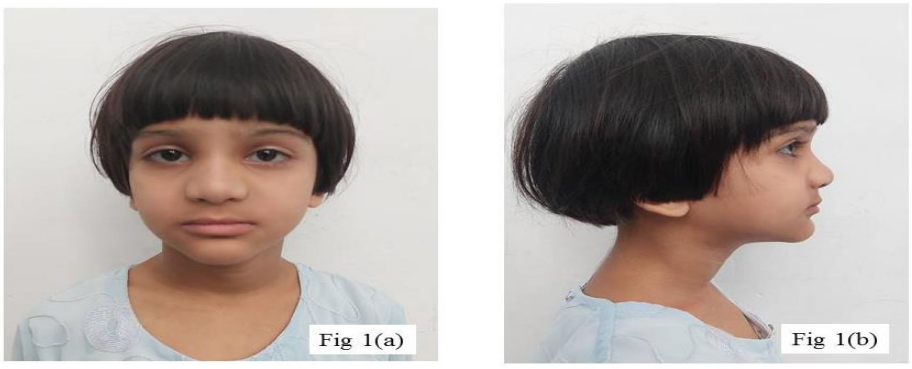


**Fig. 2: Showing Frontal and Lateral view of face.**


**Fig. 3(a): Shows polydactyly of both hands, sausage-shaped digits and clinodactyly, Fig 3(b) shows sausage-shaped digits, clinodactyly and hypoplastic nails of feet, Fig 3(c) showing hypoplastic nails of toe, Fig 3(d) showing thin and friable hair, scalp showing fungal infections.**

Intraoral examination revealed absence of mandibular anterior mucobuccal fold with hyperplastic frenula. Notching of gingiva in upper and lower anterior region was present and patient had a deep palatal vault. 4 lower incisors along with upper laterals and left central incisor was found to be missing clinically. Primary molars were carious in both maxillary and mandibular arch. Serrated incisal margin of right primary central was also noticed, along with gingival recession in mandibular Left and Right first molars.

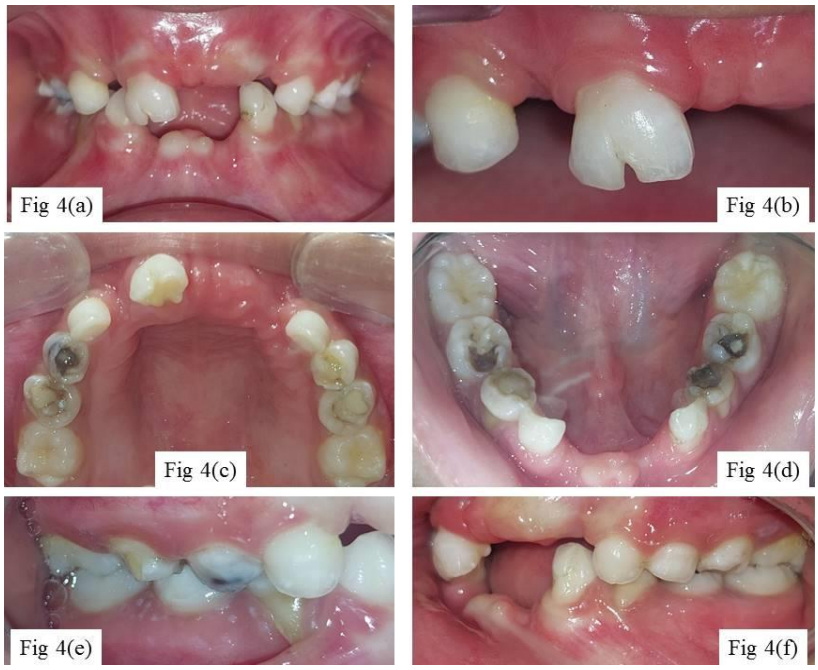


**Fig. 4(a): Shows intraoral photograph in frontal view, Fig 4(b) showing serrated incisal margin of 51, Fig 4(c) and 4(d) showing maxillary and mandibular occlusal view, Fig 4(e) and 4(f) shows posterior teeth in occlusion.**

An orthopantamogram was taken, revealing congenitally missing 12, 22, 31, 41, 32 and 42 (fig 5a). Talons cusp was visible in relation to 11 and 21 (fig 5b). An X-ray of hands and feet was also taken (fig 5c, 5d).

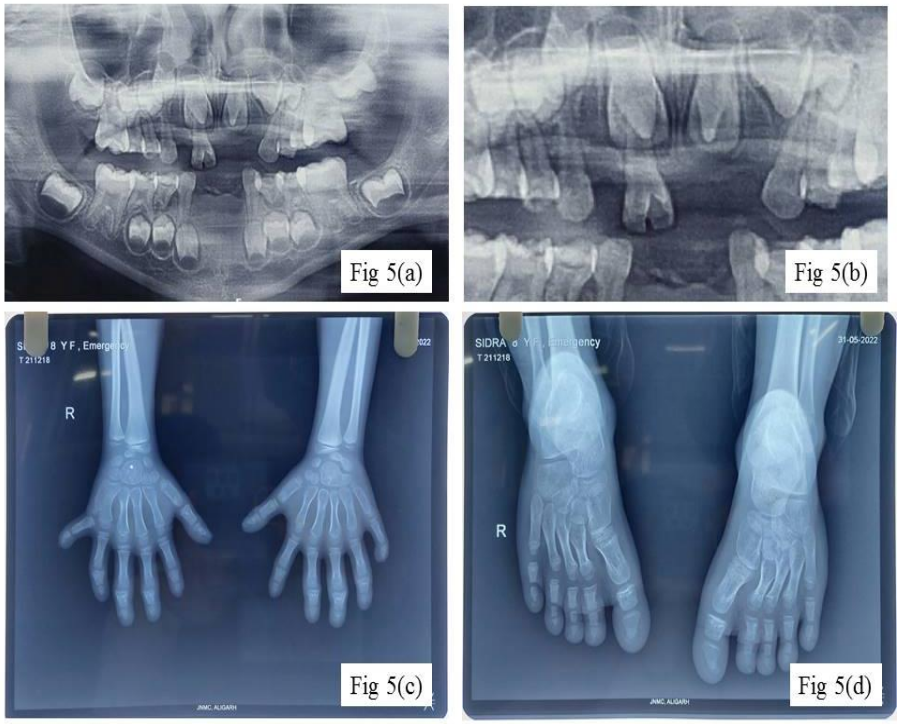


**Fig. 5(a): OPG showing multiple missing teeth, Fig 5(b) showing Talon cusps of 11 and 21, Fig 5(c) and 5(d) showing radiographs of hands and feet.**

Treatment plan for this patient include maintainence of oral hygiene, restoration of decayed teeth, replacement of congenitally missing anteriors using removable partial denture, and later by implants, referral to pediatrics, regular recall and follow up.

Oral prophylaxis and restoration of decayed teeth were performed. However, the patient was not willing for extraction and replacement of teeth by removable appliance. Hence, patient is kept on regular follow up.

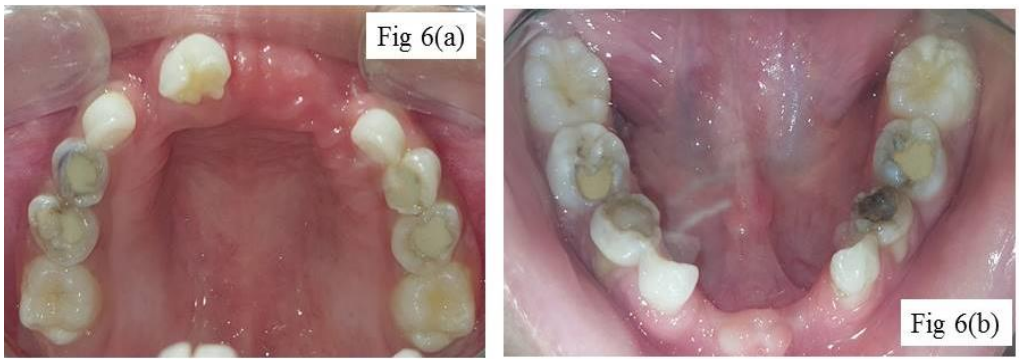


**Figure 6(a) and 6(b): Shows restored teeth.**

## DISCUSSION

Early childhood is where many disorders can be identified, and children with these conditions need special care from birth. One of these diseases, Ellis-van Creveld syndrome, is characterised by chondrodysplasia and ectodermal abnormalities.[7] From the 18th week of pregnancy onward, EVC can be identified by ultrasound in the prenatal period and by clinical examination in the postnatal period (by a tetrad of symptoms: chondroectodermal dysplasia, polydactyly, cardiac abnormalities, and congenital dental hypoplasia).[8]

The bones are affected by ectodermal dysplasia, which results in a significant skeletal ossification deficiency, shortening of the ribs and limbs, and pathological fractures. The patient frequently exhibits a narrow chest, lumbar lordosis, valgum genu, thinning hair, and an excavation known as a funnel chest.[9] Polydactyly can be seen in the hands and feet, usually there will be changes in their shape associated with brachydactyly, clinodactyly, gap between big toe and other two toes, small phalanges and sausage-shaped fingers. Fingernails are usually hypoplastic, dystrophic and friable.[10] The present case also had polydactyly of both hands, sausage-shaped digits and clinodactyly. Lumbar lordosis was also noticed.

Congenital cardiac malformations are found in 50–60% patients affected by this syndrome, and they affect mitral valve, tricuspid valve, and the atrial and ventricular septa and are responsible for the decrease in life expectancy in these patients.[11] It is usually associated to congenital heart malformations, thoracic cage abnormalities, respiratory insufficiency, and an increased risk of pulmonary hypertension. Renal disease or small kidneys, liver disease and micropenis are also frequent findings.[12]

Ellis-van Creveld syndrome presents diverse oral manifestations of the soft tissues and teeth. They include hyperplastic frenula, absence of mucobuccal fold, serrations in alveolar ridge, multiple small alveolar notches, partial cleft lip, partial anodontia, neonatal teeth, peg-shaped laterals, conical and microdontic teeth, enamel hypoplasia, delayed eruption of teeth, taurodontism, high arched palate.[1,3] The present case also have absence of mandibular anterior mucobuccal fold with hyperplastic frenula, notching of gingiva in upper and lower anterior region. 4 lower incisors along with upper laterals were found to be missing.

Malocclusions are commonly seen in Ellis-van Creveld but are not of any specific type. It has been described that hypoplasia of the anterior maxilla, prognathism of the mandible and increased height of the lower third of the face are common finding noticed.[13]

50% of the children with Ellis-van Creveld syndrome die in early infancy due to cardiorespiratory problems. Those children who survived require a multidisciplinary approach for treatment, i.e. orthopedic correction of genuvalgum, amputation of extra digit, surgical repair of cardiac malformation and dental intervention.[14]

Treatment planning for Ellis-van Creveld syndrome may require an interdisciplinary approach. Treatment needs include preventive measures against caries, restorations of decayed and malformed teeth, partial dentures with frequent adjustment and replacement and correction of malocclusion. Children with Ellis-van Creveld syndrome have a high risk for caries because of malformed hypoplastic teeth and molars with deeps pits and fissures. Therefore, importance of home-based preventive measures such as diet control, oral hygiene maintenance, daily fluoride mouth rinses and use of fluoridated denitrifies should be made aware to the patient and the patient's parents. Preventive professional care such as pit and fissure sealants, oral prophylaxis and professional topical fluoride application must also be provided.[15]

Congenitally missing teeth, hypoplastic teeth and malocclusion can affect speech, appearance and

mastication of these patients. To maintain space and to improve speech, mastication and esthetics, partial or fixed denture is advocated. Restorations of hypoplastic and decayed teeth with composite are done for better esthetics and preservation of tooth structure.[1] Parental and child counseling is required for psychological trauma due to compromised oral manifestations and medical health. Surgical correction is advised for soft tissue and skeletal abnormalities.[3]

Antibiotics prophylaxis is recommended for surgical patients with cardiac defects to prevent infective endocarditis before invasive dental procedures.[16]

## CONCLUSION

Ellis-van Creveld syndrome is an autosomal recessive disease with a characteristic tetrad of chondrodysplasia, polydactyly, ectodermal dysplasia, congenital heart malformation. Recognizing the oral findings is important for early diagnosis and a correct treatment planning, and in this aspect dentists may play a vital role. To achieve satisfactory functional and aesthetic results, multidisciplinary treatment planning is necessary for preventing oral disease and to provide restorative care. Collaboration between pedodontist, orthodontist, oral and maxillofacial surgeon and prosthodontist is the key to successful management.

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