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Ellis–Van Creveld Syndrome With Unusal Expression Of Multiple Supernumerary Teeth : A Case Report

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Abstract: *Ellis–van creveld syndrome is a rare autosomal recessive syndrome of the heart, skeleton and most of the ectodermal derivatives. It is caused due to genetic defects in the chromosome 4p16; it belongs to short rib-polydactyly group (SRP). It is characterized by short ribs, short limb, polydactyly, ectodermal dysplasia and congenital heart defects. The oral manifestation is one of the main characteristic diagnostic features and it includes partial anodontia, microdontia, fusion of upper lip to the gingival margin, multiple frenum, anterior open bite etc. We the dentist play a major role in diagnosing such patients, managing their dental problem and help them lead a normal life. We report a case of EVC syndrome expressing all the findings with multiple supernumerary teeth.*

Key words: *polydactyly, multiple frenum, genu valgum, multiple supernumerary teeth, partial anodontia.*

I. Introduction :

Ellis-van Creveld (EVC) syndrome is a congenital genetic disorder common among the Amish population (1 in 5000 live birth), in general the worldwide prevalence rate is 7/1,000,000 live births.[1] No data is available for prevalence rate in the Indian scenario but Approximately less than 25 cases only have been properly reported in India till date.[2] Baujat G *et al*. described that about 150 cases were reported between 1940 and 2008.[3]

In 1940, Richard W. B. Ellis and Simon Von Creveld described Ellis Van Creveld (EVC) syndrome. EVC is also termed as chondro ectodermal dysplasia or meso-ectodermal dysplasia[4] EVC is an autosomal recessive congenital genetic disorder[4], occuring due to the genetic defect located in chromosome 4p16.[4] Mutation of EVC1 and EVC2 genes, located in a head to head configuration on chromosome 4p16 has been identified as causative focus. They occur due to genetic defect in initiation and morphodifferentiation stage of tooth development.[6]

II. Case Report

A 21 year old male patient reported to us with a chief complaint of missing upper front teeth and malposition of lower right and left back teeth since 10 yrs of age. Patient history revealed that he had a similar complaint with his deciduous teeth which was neglected by his parents. The past medical history suggested that, at the age of 4 he was a diagnosed with Atrial septal defect and was treated for the same. Patient also gives history of noticing stunted growth and deformity in his hands and feet, since his childhood for which he did not seek any medical opinion. Presently the patient reported to us for reasons of social stigma and esthetic concerns. Family history suggested that he was born to parents of consanguineous marriage and he was their only son. There was no history of similar complaints in his family.

On general physical examination the patient had a short stature 145 cm and weighed 67kg (Fig 1). Shortness of the limbs, genu valgum deformity (Fig 3) and polydactyly noted in both the hands (Fig 2). The hands and feet were wide and markedly deformed, sausage-shaped fingers and dysplastic fingernails were present. No dryness of skin was noticed and patient sweat was normal, no quantitative change in hair noticed, however there was focal hypotrichosis of the eyebrows and mild alopecia of the left hair line region was noticed.

Extra oral examination revealed broad nasal bridge, no facial gross asymmetry present, TMJ movements are with in normal limits, and no abnormality detected in submandibular and parotid region, no palpable regional lymph nodes was present.

On intra oral examination thick labial frenula in relation to 71,81,14,13 region and multiple accessory frenulum's were present in maxillary and mandibular labial sulcus regions. Supernumeries in relation to 15, 25, 32, 34, 35, 42, 43, 44, 45; fused 23, Screw driver shaped 21 with talons cusp on labial surface noted. Patient also presented with anterior open bite, grade II mobility in relation to 32, 42, 43 and generalized probing pocket depth of 4 to 5 mm was present (Fig 4). Correlating it with the history and the clinical features, a provisional diagnosis of Ellis van Creveld syndrome was arrived. Then the patient was subjected to routine blood investigations, ECG, ECHO and orthopantomograph (OPG).

Routine blood investigations suggested that all the blood values were within normal limits. ECG was normal and ECHO showed normal study. OPG (fig 5)showed prominence of the antigonial notch bilaterally, decreased anteroposterior width of the mandibular ramus bilaterally missing maxillary and mandibular anteriors, multiple retained deciduous teeth. Multiple Supernumery teeth present in relation to 14, 15, 24, 25, 26, 34, 35, 36, 44, 45, 46 region (Fig 4).

Considering the nature of the condition, the patient/parents were cautioned about the possible complications and advised periodic systemic review. With regard to dental rehabilitation the patient was scheduled for extraction of retained deciduous and supernumery teeth. Followed by frenectomy, orthodontic correction and prosthodontics replacement of missing teeth.

III. Discussion

EVC syndrome belongs to short rib polydactyly group SRP III - Verma- Naumoff syndrome.[4] EVC syndrome is recently included in new class of human genetic disorders called as ciliopathies, which is due to defective molecular mechanism of primary cilia.[7] EVC syndrome can be diagnosed prenatally from the 18th week of gestation, by ultrasonography / fetoscopy[8] or by clinical examination after birth.[3,4,9,]

According to Arumugam aashish et Al , child born to EVC patient have a chance of transmitting its autosomal recessive gene is 25%.[10] Ellis–van creveld syndrome is common among Amish population of Pennsylvania in USA[4]. History of parental consanguinity is very common in EVC syndrome patients[5,7,], similar to our patient. Average Adult height for EVC syndrome patient is 109 to 165 cm[1,2]

The characteristic features of EVC syndrome is the presence of dysplastic nails and typical deformation of fingers- polydactyly, short ribs, short limbs, genu valgum deformity[1,4] ,Congenital heart malformation(60% of cases), which is commonly a single atrium. Basically the main determinants of longevity in EVC syndrome are the presence of congenital heart disease and thoracic dysplasia or respiratory distress. [2,4,]

According to Kapoor et al,[5] the tetrad of cardinal features seen in EVC syndrome is:-
(1) Disproportionate small stature and shortening of the middle and distal phalanges;
(2) Polydactyly affecting hands;
(3) Ectodermal dysplasia mainly affecting nails and teeth;
(4) Congenital heart malformation.

All the above mentioned tetrad of cardinal sign was clearly present in our patient

The oral manifestation is one of the main characteristic diagnostic features, the oral findings includes natal teeth, conical shaped or missing teeth, premature eruption, and/or exfoliation of teeth and absence of labial vestibule because of fusion of upper lip to the gingival margin, leading to the notching of upper lip.[3,5,] These features were also present in our case; in addition had a talons cusp in 21. Anterior open bite was also present in our case. The oral findings could be due to the continuation of the normal serrated condition of the gingiva from the third to seventh month in the uterine life of the fetus and it affects the esthetic problem, speech, jaw growth of the child.

According to Kar et al, the most common Differential diagnosis for EVC syndrome includes Jeune syndrome and Orofaciodigital syndrome. Jeune syndrome is autosomal recessive disease; characterized by thoracic dystrophy, short limbs, small stature, polydactyly and generalized bony dysplasia. Eye, kidney and lungs are also affected. The orofaciodigital syndromes result from dominant sex-linked inheritance and are limited to women. This condition is clinically characterized by multiple gingivolabial frenula, hypoplasia of the nasal cartilages, moderate mental retardation and fissured tongue. Ankyloglossia is an important feature and it is found in one third of cases.[9]

The management of EVC is usually symptomatic and it is accomplished with multidisciplinary approach. Patients generally need the consultation of Psychologist, pediatrician, pediatric neurologist, clinical geneticist, Pulmonologist, Cardiologist and Physiotherapist. For rehabiliatation plastic surgeon, stomatologist, endodontist, general Dentist, oral & maxillofacial surgeon, prosthodontist and orthopaedician play a vital role.[3,4,.]

IV. Conclusion

Oral manifestation is one of the characteristics of EVC syndrome. Oral changes can affect esthetics, speech and jaw growth causing a social stigma, as seen in the present case. Thus dentists should play a lead role in early diagnosis and establishing treatment protocols that improve the aesthetics, functional efficiency and quality of life.

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Fig 1: front and side profile

Fig 2: genu valgum

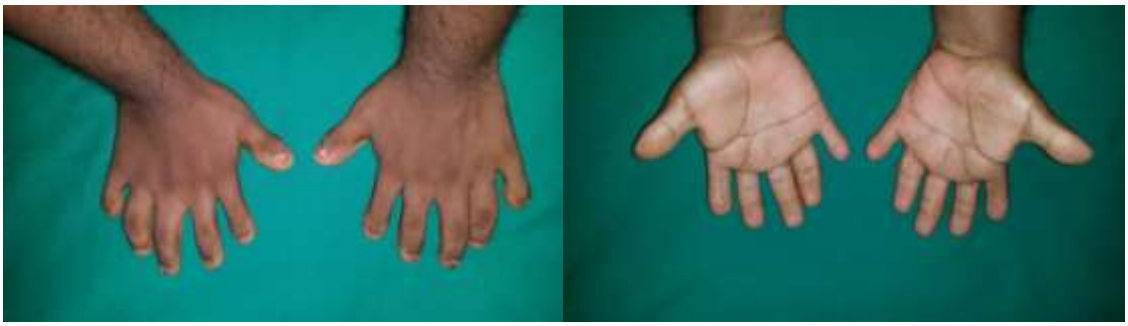

Fig 3: polydactyly – dorsal and ventral surface of palm

Fig 4: intra oral photograph: multiple frenums, thick labial frenum, partial anadontia , multiple supernumerary teeth

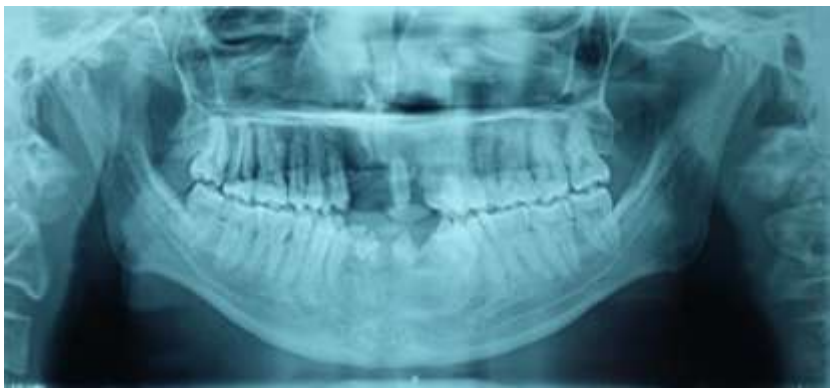

Fig 5: orthopantamograph

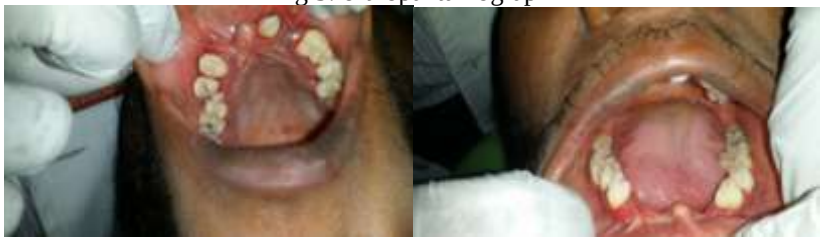

Fig 6: post-operative photograph after oral prophylaxis and dental extraction .