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Oral and facial features in Ellis-van Creveld syndrome: A case report

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Abstract

The Ellis Van Creveld syndrome is chondroectodermal dysplasia form, which is characterized by skeletal abnormalities and ectodermal derivatives.

The clinical manifestation observed, in particular oral and facial manifestation will be described in this syndrome. The abnormalities recognized in 8 years old boy who carries a congenital heart disease.

Besides the numerous decay, oral manifestations are dominated by oligodontia, the presence of multiple frenulum and flanges.

The orthopedic abnormality suggestive of an achondrodysplasia well as skin lesions led us to seek a syndromic reached.

The multidisciplinary approach is early in the specialties of pediatric surgery and cardiovascular surgery. The fact remains that the Stomatological management also needs, the contribution of several dental services is essential to treat oral lesions and prevent their complications.

Keywords: Ellis-van Creveld Syndrome, chondroectodermal dysplasia, oligodontia, polydactyly, congenital heart disease.

Introduction

The syndrome Ellis Van Creveld of (EVC) is a rare chondroectodermal dysplasia with 7/1000 000 births prevalence, characterized by skeletal abnormalities and ectodermal derivatives.

It was described for the first time by Richard B Ellis Edinburgh and Simon Van Creveld Amsterdam in 1940 [1].

The syndrome is identified by MIM ID # 225500 [2] and characterized by the tetrad: disproportionate dwarfism, dolydactyly, ectodermal dysplasia, congenital heart disease.

The pathophysiology of this syndrome is unknown, but studies show that it has a genetic carrier whose scientific advances tend to elucidate [3-5].

Histopathologically, observations revealed that the cartilage of the long bones is the site of a disorganization of chondrocytes in the growth zone [6]. Orofacial anomalies derived from the originaldevelopment of ectodermal defects. Oral manifestations in the EVC syndrome are characteristic and constant. Dentists must know the clinical features of Ellis Van Creveld syndrome and complications that can occur during this genetic disease.

The goal of the work is to describe through observation, orofacial manifestations and to discuss the therapeutic considerations.

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Observation

An eight old boy was referred to the dental and oral surgery by his dentist for multiple tooth extractions on a heart field.

The patient's medical history was notable for congenital heart disease with Atrio Ventricular Communication (AVC) was operated at the age of five years. Currently the patient has recovered and does not undertake any medical treatment.

The child has been operated in pediatric surgery for polydactyly of the both hands at the age of six years. He was born of consanguineous parents of 2nd degree, parents are phenotypically healthy.

Mother 38 years old, had five pregnancies with two deaths at birth where the children had malformations, polydactyly and heart problems. The boy was born prematurely at 34 weeks with respiratory distress. He weighed 3.5Kg and measured 47.5cm. It is the latest in a family of 3 children who do not show signs. The diagnosis was prenatal malformations.

Motor development and teething took place in a normal time. Currently, the child is followed in the pediatric ward, for a failure to thrive, it is taking growth hormones. The child has normal schooling and won high marks.

A general examination is actually notices the stature, with a disproportionate dwarfism: a narrow thorax, legs and short arms to the torso and genu valgus, 112cm measuring and 22kg weighing.

In hands found the scar made extra finger, as hypoplastic nails of fingers and toes (fig.1,2).
A facial review include sparse hair (fig.3.), a broad forehead, an unverted nose with flat nasal bridge (fig.4.), short upper lip contrasting with the lip lower, forked philtrum (fig.5.), a tendency to class III.

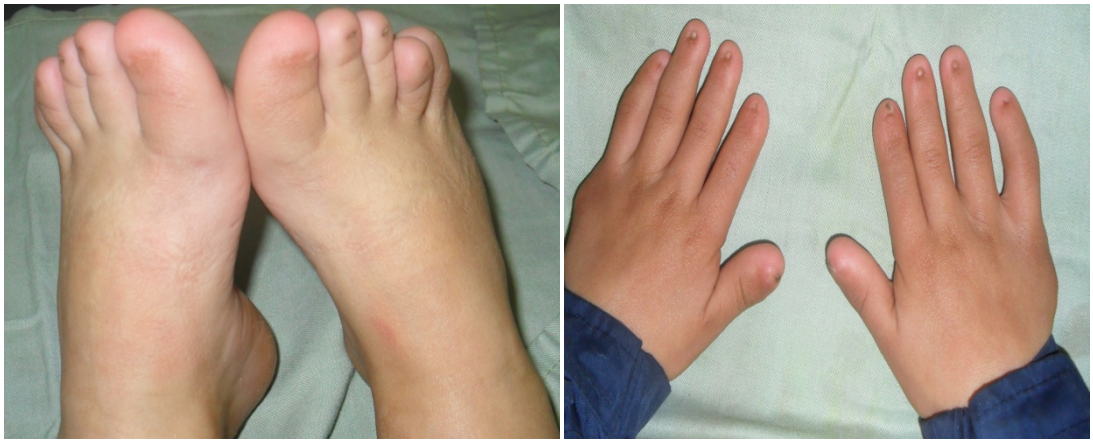
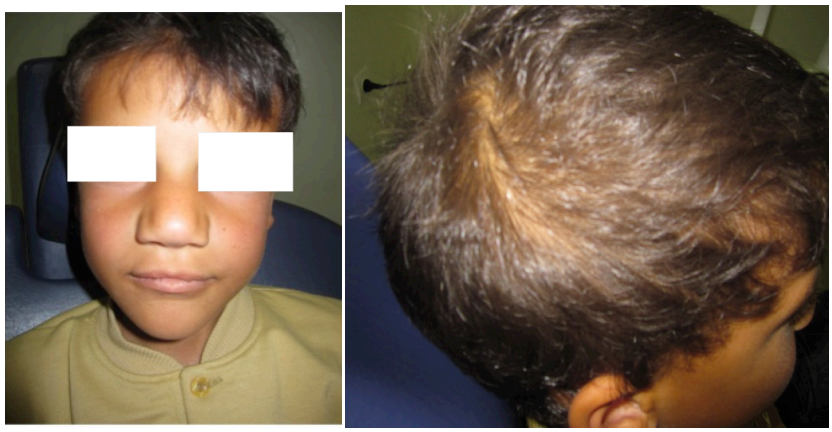


Figure1, 2 : The patient's Toenails and fingernails were small and dystrophic The ulnar aspect of both hands demonstrated the surgical repair of polydactyly



Figures 3, 4: broad forehead, unverted nose with flat nasal bridge Sparse thin hair

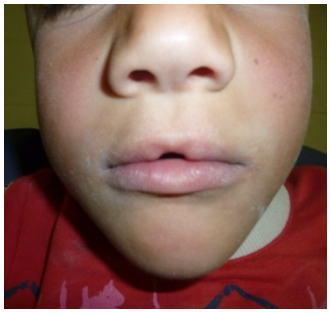


Figure 5: the short upper lip presents a slight V-notch in the middle due to fusion of the anterior portion of the upper lip to the maxillary gingival margin.

All these characteristics confer to have likeness between all patients EVC syndrome.
The oral examination reveals poor hygiene, with a strong predisposition to caries, the DMF index is 18 and oligodontia of the primary toothing. At first glance, the dental caries is deep, but all teeth respond to vitality tests.

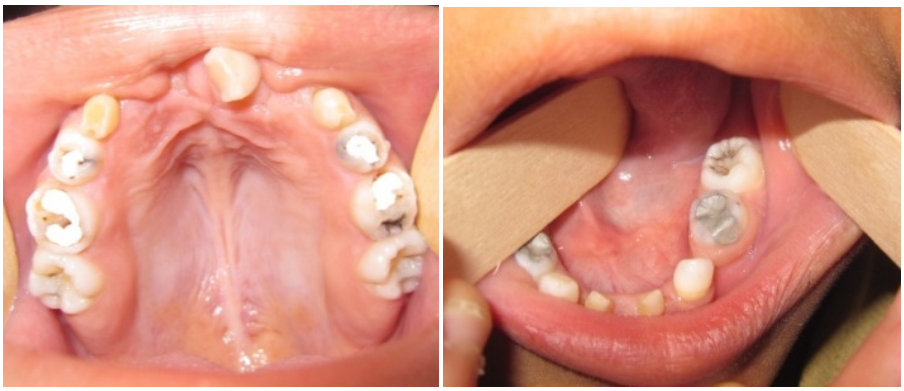


Figure 6, 7: Deep palate with protruding intermaxillary suture in anterior portion, first lower left molar with abnormal cusps or accessory grooves

The 61, 72, 82, are atypical, conical microdontia, with enamel hypoplasia. The 36 has an abnormality of the cusp slopes with many craggy grooves (fig.6, 7). The upper and lower frenum have a wide insertion with the presence of numerous secondary brakes. The vestibule is nonexistent by fusing the inner face of the lip with the gum (Fig.8, 9); but the tongue is not stiff. Furthermore, the palate is deep, covered by a thick fibromucosa with hand clamps and other intermaxillary suture protruding at the anterior tiers (Fig.6).
Examination of the occlusion reveals an open bite and class III right and left.

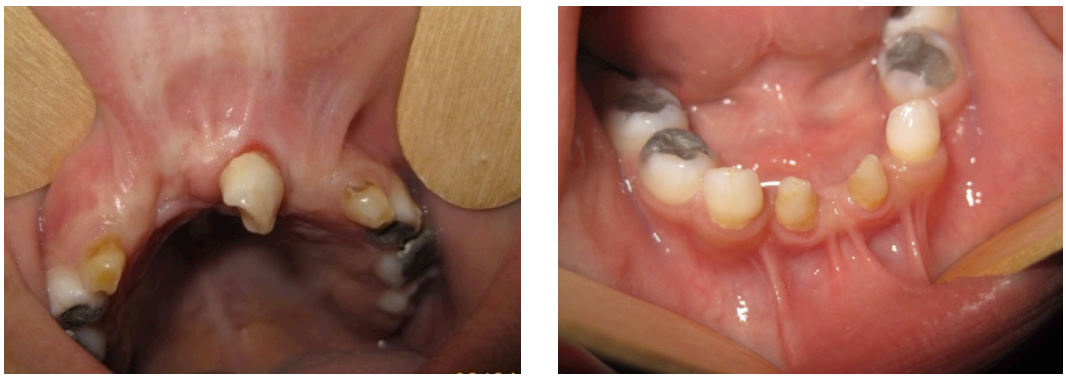


Figure 8, 9: fusion of the upper lip to the gingival margin, microdontic, missing and cone-shaped teeth. The anterior labiogingival sulcus obliterated by multiple frenula, frenulum hypertrophy.

Panoramic radiography (Fig.10) confirms the presence of germs of 13, 23, 14, 24, 15, 25, 17, 27, 33, 34, 35, 37, 44, 47. The 11 and 21 are atypical. Maturation 6 and 7 is compatible with the civil age: eight years. There are also no germs of 22, 12, 32, 31, 41, 42, 43 and 45.
A discreet taurodontism on 85, 75 and first upper right molar. In total there are 15 temporary teeth and 28 permanent teeth.

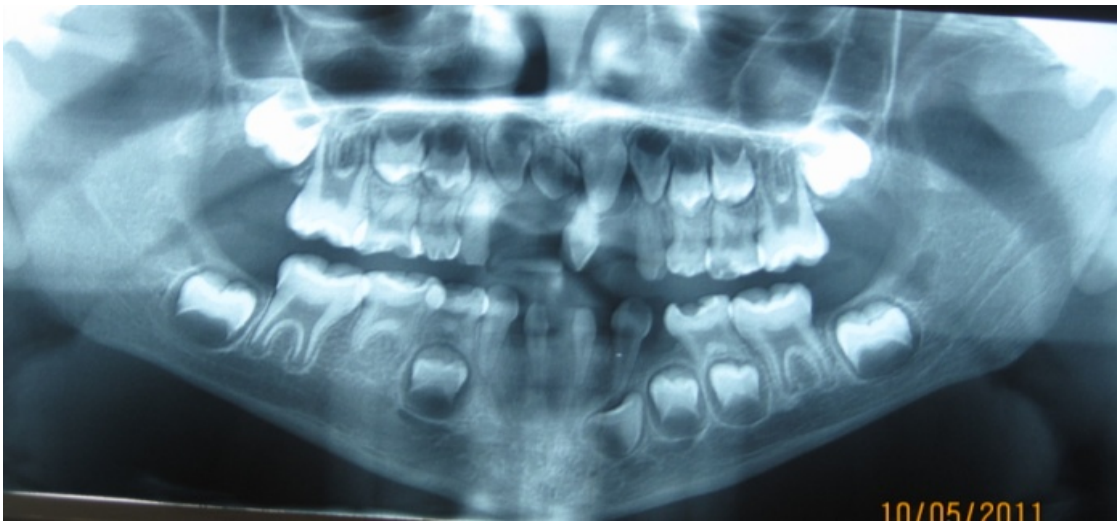


Figure 10: congenitaly missing of permanent teeth (12,22,32,31,41,42,43,45) taurodontism (75, 85and 16)

On the radiograph of the hands (fig.11), bone age is 7-8 years depending on the stage of Greulich and Pyle. There are also symmetrical involvement: hexadactyly post-axial, hypoplasia of the terminal phalanges, an incomplete duplication or fusion (synostosis) of 5th and 6th metacarpal. Radio foot (fig.12.) Include curved bone, a valgus knee, and humerus deformed cross, the epiphyses are widened.
The mail cardiologist indicating that cardiac involvement in unique type of headset as part of a partial AVC confirmed by Echo-Doppler heart and the right heart catheterization.
Congenital heart disease was operated at the age of one year and then return at the age of five years following the dropping of the patch. The long-term development will depend on the holding of the currently satisfactory mitral valve repair.

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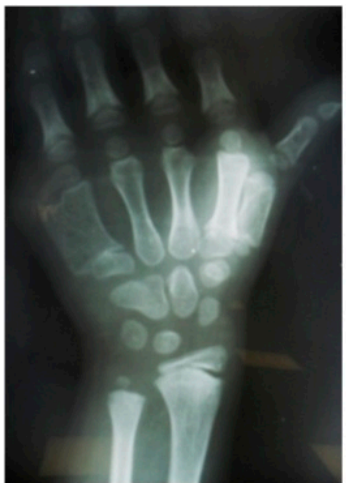

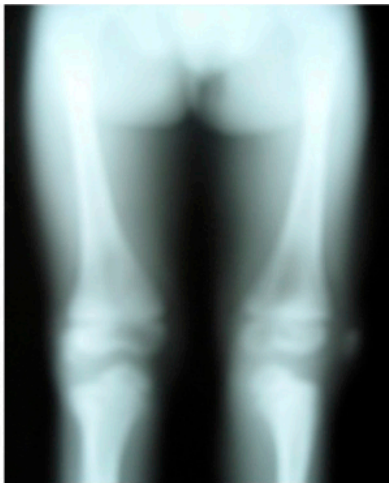

Figure 11, 12: incomplete duplication of the 6th metacarpal. X-ray of knee showing genu valgus , deformity of humerus and widened epiphyses

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The risk of infective endocarditis being major, prophylactic antibiotics Amoxicillin or ampicillin 75 mg/kg P.O. or I.M. or I.V, if the patient is allergic to penicillin/amoxicillin the use of Clindamycin 20 mg/kg P.O. or I.V. Cefalexin or cefadroxil 50 mg/kg P.O. Cefazolin 25 mg/kg I.M. or I.V. Azithromycin or clarithromycin 15 mg/kg P.O. were recommended one hour before a bloody act as a tooth extraction. The diagnosis of dentinitis was covered all primary teeth caries. A treatment plan was established and appliqued containing**:** First the motivation to oral hygiene and explanation of the brushing method and information for parents on the need for a rigorous oral hygiene, as well as a sugar consumption decrease, given the reduction of dental capital. Then conservative treatment of primary teeth decayed by an amalgam restoration, composite or by pedodontic caps and the maintain of permanent teeth: 16-26-36-46 by a sealant, and a topical fluoridation with restoration of prosthetic transition to the upper incisor with a crown on 61 conoid, which will allow the aesthetic rehabilitation and also serve as a space maintainer.

Also monitoring of the physiological establishment of permanent teeth and bearing the edentulous (anterior mandibular sector) is a removable prosthetic restoration, and providing periodontal surgery such as frenotomy and deepening the vestibule or prosthetic referred. An Orthopedic treatment of Class III was need, so the patient was referred to an orthodontist.

Discussion

The syndrome Ellis Van Creveld of dysplasia or ectodermal chondro is a rare disease: 7/1000 000 births prevalence. So far, most cases about 300 were reported through literature, in all countries of the world where events were described by highlighting injuries as specialties [7-13].

Transmitted in an autosomal recessive mode, the location is on the 4p16 gene, there is a mutation of the gene at 2 levels: EVC1 and EVC2 [3-5].

The concept of inbreeding found in 30% of cases, increases by 50% the risk. In Algeria, the high prevalence of consanguineous marriages and the many siblings explain the high number of observations of EVC syndrome. Better known in pediatrics and pediatric surgery, orofacial manifestations are also very evocative.

The skeleton of the examination is still necessary to define the anomalies. Signs are the acromésomélie. Polydactyly postaxial is a constant feature. Multiple skeletal abnormalities are described, such as the merger of carp, small iliac crest, valgus knee, fibula disproportionate to the tibia, chest and narrow ribs [14, 15]. The disproportionate dwarfism affecting the lower limbs (mesomelic part), is associated with growth retardation. Hormone treatment is controversial. The therapeutic means are moving towards the surgical extension appears to be a better method [16].

Dental abnormalities dominate the clinical picture of oral manifestations.

• They are number where agenesis is a cardinal point primarily in the anterior region [17-19], but as paradoxical as it seems, the natal or neonatal tooth is a supernumerary tooth described in 25% of cases. This anomaly is not found in our patient.

• Structure abnormalities in type hypomineralization enamel

• Shape abnormalities manifest themselves in conoids teeth, taurodontism or talon cusp.

• Molars having abnormal accessory cusps or grooves with deep pits and fissures which explains the observed caries. [20]

Susceptibility to caries is also due to associated diseases requiring hospitalization, spreading brushing and the many measures taken drug (syrup at bedtime). In our case we describe many dental abnormalities that are similar to all those described in literature and occurring with this syndrome.

Congenital heart defects occur in approximately 50-60% of case sof this syndrome. The most common is atrial septal defect (atrioventricular canal: AVC) [21], that defect was also found in our case. Other heart defects are ventricular septal defect, hypoplasia of the aorta and the only headset that requires early surgical repair. These diseases are responsible for respiratory distress and neonatal mortality.

Prenatal diagnosis is possible from the 18th week of gestation by an ultrasound that can detect heart defects, kidney, chest and the member's developmental defects [22-24].

Differential Diagnosis discusses several syndromes that cause dwarfism and polydactyly.

• Facial oro digital syndromes (OFD) [25, 26] comprise an heterogeneous group of disorders characterized by facial and oral digital anomalies. They have sub groups of from 1 to 11 and type V is represented by Thurston syndrome [27, 28],

• The achondrodysplasia is easily eliminated because the phaneres, the extremities and the teeth do not show abnormalities.

Support for dentistry, including bloody acts such as dental extractions and frenectomies be done under antibiotic prophylaxis of bacterial endocarditis.

Conclusion

Dental and orofacial multiple defects that characterize the Ellis Van Creveld syndrome make the diagnosis suggested earlier by the dentist at a young age of the patient. The diagnosis can be confirmed by examination of the hands remain available to the research review digital anomalies and the dentist can verify the existence of an associated congenital heart disease. This approach allows for optimal support that fits into the context of multidisciplinary cooperation.

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