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Case Series

The Role of The Pediatric Dentist in the Diagnosis and Management of Ellis Van Creveld Syndrome

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Abstract

Ellis-van Creveld syndrome (EVC) is a rare autosomal recessive disorder caused by mutations in both the EVC1 and EVC2 genes, with a birth prevalence estimated at 7 per 1,000,000 in the general population. Parental inbreeding has been reported in 30% of cases. The syndrome is characterized by chondral and ectodermal dysplasia, leading to features such as short ribs, polydactyly, stunting resulting in dwarfism, dental and craniofacial abnormalities, and cardiac malformations. In two reported cases, a 5-year-old girl and a 9-month-old boy exhibited general and oral manifestations typical of EVC syndrome. Accurate diagnosis during the first months of life is crucial for the overall prognosis of the syndrome. Regular clinical follow-up is essential to prevent complications and to ensure timely management.

Keywords: Autosomal Recessive; Multiple Dental Agenesis; Heart Defects; Polydactyly

Introduction

Ellis-van Creveld syndrome, also known as chondroectodermal dysplasia, is an uncommon autosomal recessive skeletal dysplasia characterized by polydactyly, ectodermal dysplasia, chondrodysplasia and congenital heart defects . In 1964, McKusick et al. reported 52 cases of CVS in the Old Amish population, 30 of which died in the first year of life [1-3].

This syndrome was discovered by Richard WB Ellis and Simon van Creveld in 1940 [4]. Some studies have reported a higher prevalence of the disease among amish, Brazilians, Ashkenazi Jews and Arab communities with high rates of inbred marriages. Although most patients have normal intelligence, central nervous system abnormalities or mental retardation have sometimes been reported [5].

It is caused by mutations of the EVC1and EVC2 genes and shows autosomal recessive inheritance with parental consanguinity in about 30% of cases [6].

The diagnosis of EVC syndrome is established in a proband with characteristic clinical and radiographic findings and biallelic pathogenic variants in *DYNC2H1*, *DYNC2LI1*, *EVC*, *EVC2*, *GLI* , *SMO*, or *WDR35* or a heterozygous pathogenic variant in *PRKACA* or *PRKACB* identified by molecular genetic testing [7].

Regarding oral manifestations, Ellis-van Creveld syndrome is characterized by dental abnormalities such as microdonte, conical teeth, taurodontism, supernumerary teeth, enamel hypoplasia, neonatal teeth in about 30% of cases, as well as premature exfoliation of teeth, malocclusions, absence of vestibular sulcus, hypertrophic lip brake, and dystrophic philtrum may also be observed [8].

In this work, we report two cases of children diagnosed with Ellis-van Creveld syndrome presenting all the oral manifestations described in the literature.

Clinical Cases

1st case

A 5-year-old girl was referred to the pediatric odontology department at the La Rabta hospital in Tunis accompanied by her parents who had as a reason for consultation: the absence of temporary mandibular incisors on an arch. The diagnosis of EVC was made at birth by the geneticist. There was no family history of the disease, the patient had healthy parents, she was from a first degree inbred marriage. Pregnancy and childbirth went well leading to a full-term birth. There was no history of epilepsy or neurological impairment.However, she had congenital heart disease.On general examination, she had a small dysharmonic size, short upper and lower limbs with bilateral hexadactyly and hypoplastic nails (Figure 1).

Figure 1: Rahma, aged 5, has Ellis-Van-Creveld syndrome, is referred following a delay in the eruption of her teeth: she is small with short limbs and sparse hair, polydactyly and brachydactyly of the hands, hypoplastic nails.

The patient had characteristic oral abnormalities, including hypertrophic upper lip brake with low insertion (Figure 2).

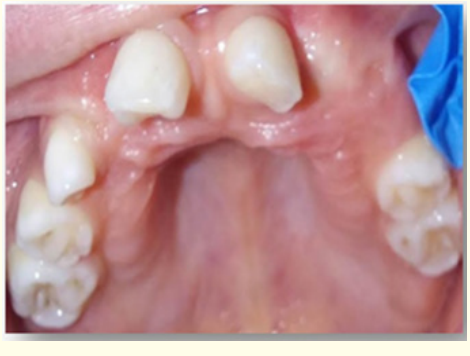

Figure 2: Low insertion of the upper labial frenulum, significant inter-incisor diastema, absence of 62, 63, 52, tooth shape anomalies.

It also presented dental abnormalities of form (Figure 3): conical and ankle-shaped teeth.

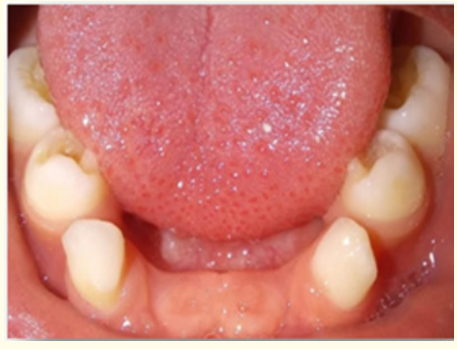

Figure 3: Absence of 81, 71, 82 and 72, shape anomalies of all the teeth.

Permanent molars with a single root, number: a hypodontia of temporary or permanent teeth, structure: a hypoplasia of enamel, microdonte, taurodontism (Figure 4) and delayed eruption of temporary teeth.

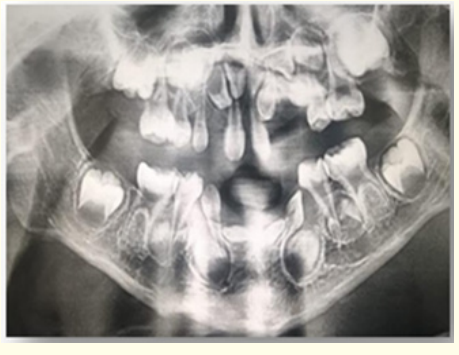

Figure 4: Anomalies of tooth germ shapes, retained germs, upper 1st molar with a single conical root, Taurodontism in the four second molars.

A lower removable partial denture was added to replace 31, 32, 41 and 42 and another to replace 52, 62 and 63 (Figure 5).

2nd case

A 2-year-old child accompanied by his parents attended our consultation. He has Ellis-van Creveld syndrome and followed in pediatric cardiology for wide interauricular communication.it was the result of a 1st degree inbred marriage.

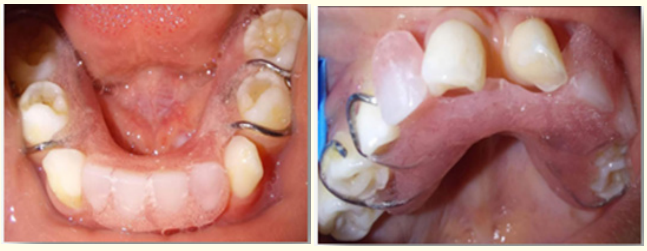

Figure 5: A removable partial prosthesis replacing 31, 32, 41 and 42, A removable partial prosthesis replacing the 52, 62, 63.

In addition, the patient is followed by an otolaryngologist for chronic nasal obstruction resulting in a marked increase in the effort required to breathe and an increase in the respiratory rate. The patient had no history of associated fever, chest pain or frontal perspiration, but suffered from severe cough and cold, which favored oral breathing. This oral breathing can cause problems such as dehydration and poor sleep quality. Chronic nasal obstruction may be due to nasal malformations or enlarged tonsils or adenoids.

At the general examination level, the patient has a macrocephaly. He also has polydactyly (Figure 6).

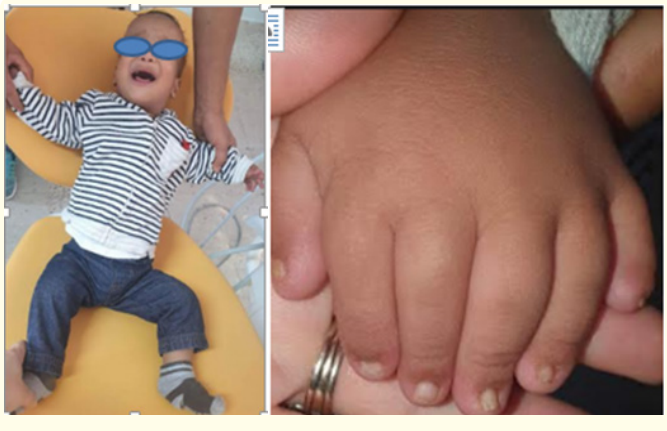

Figure 6: Ahmed , 2 years -old, has Ellis-Van-Creveld syndrome, is referred following a delay in the eruption of his teeth: small with short limbs and sparse hair, polydactyly and brachydactyly of the hands, hypoplastic nails.

The disproportion between different body parts is also present with short limbs and a larger trunk.

During the endobuccal examination, the upper incisivo-canine group (Figure 7) and lower group with an ogival palate were missing.

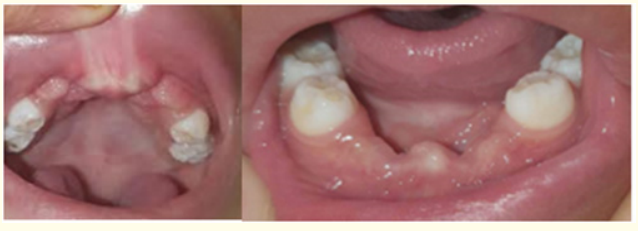

Figure 7: Absence of 51, 52,53,61,62 and 63, shape anomalies of all the teeth,absence of 71, 72,73,81,82 and 83. The four temporary molars of abnormal shapes with shorter and more rounded cusps than normal.

Discussions

EVC is a genetic disease that is transmitted in an autosomal recessive mode. It is most often described in families with a history of inbreeding, which has been verified in both our cases [3].

Diagnosis of this syndrome is possible in the fetus from week 18 of pregnancy by ultrasound [9].

EVC, or Ellis-van Creveld syndrome, presents with a constellation of characteristic features including chondro-ectodermal dysplasia, polydactyly, congenital heart defects, and tooth and nail hypoplasia. The prognosis of affected individuals is primarily determined by the severity of associated cardiac anomalies. Differential diagnosis is crucial, particularly to distinguish EVC from other chondrodystrophies like Achondroplasia and Jeune syndrome [10].

The spectrum of oral manifestations varies widely from malocclusion to dental and gingival problems. Delayed eruption has been reported as the most common finding [11].

The dental management of medically compromised and syndromic children poses significant challenges for oral health care providers. Syndromes such as EVC, characterized by a variable phenotype affecting multiple organs, require special attention from birth onwards. Early identification of such syndromes is critical for providing tailored dental care to ensure optimal oral health outcomes [12].

The minimum diagnostic criteria for CVS include postaxial polydactyly of the hands, and less frequently of the feet. One can also observe dwarfism, short limbs, dysplasia of nails and teeth.

The oral manifestations of EVC syndrome can help to establish a diagnosis at birth or in the first months of life, although these are numerous and involve not only soft tissues, but also the number (microdonties), the form (conical teeth) and tooth structure.

Common features that can be found in the majority of CVS patients include conical teeth, agenesis, high rate of caries and enamel hypoplasia.

The orofacial manifestations observed in our patients are consistent with those described in the literature. It has a hypertrophic upper lip brake, agenesis of some teeth, conical teeth with microdontia, enamel hypoplasia and taurodontism.

Dentists and especially pedodontists play a crucial role in the detection of oral manifestations of this syndrome in the first years of life.

The management of these syndromic patients includes several key elements, including diet control, maintaining good oral hygiene, anddental, daily use of fluoridated mouthwashes and age-appropriate fluoridated toothpastes to prevent carious diseases knowing that they are patients with congenital heart disease at very high risk of infectious endocarditis. Preventive treatments such as sealing of wells and cracks and a professional topical application of fluoride should also be provided from the eruption of 6-year-old teeth. It is necessary to carry out conservative care while taking into account the infectious risk in relation to CVS-related heart diseases, to propose removable partial prostheses to compensate for multiple agenesis with frequent adjustments and replacements, as well as correcting malocclusions. In addition, to maintain space and improve phonation, chewing and aesthetics, removable or fixed partial dentures can be offered.

Conclusion

Ellis-van Creveld syndrome requires therapeutic planning multidisciplinary. The pedodontist can play a fundamental role in the diagnosis and prevention of caries and prosthetic management of multiple agenesis.

Bibliography

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