

Chondroectodermal dysplasia (Ellis-van Creveld syndrome): a case report

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Abstract: Ellis-van Creveld (EVC) syndrome is a form of skeletal and chondroectodermal dysplasia, occurring with and without systemic involvement. Taurodontism of permanent and primary molars and upper posterior supernumerary teeth are rarely associated with this syndrome. A 5-year-old girl presented with early childhood caries and hypodontia. She had labiogingival adhesion, labiogingival frenulum hypertrophy, accessory labiogingival frenula, and a serrated appearance of the gingiva. She was also short in stature. All major features of chondroectodermal dysplasia were present. EVC syndrome requires multidisciplinary therapeutic planning, and the dentist plays a fundamental role in management of the oral and dental manifestations. (J Oral Sci 52, 333-336, 2010)

Keywords: Ellis-van Creveld; chondroectodermal dysplasia; case report.

Introduction

Ellis-van Creveld syndrome (EVC, OMIM #225500) is a chondroectodermal dysplasia with a birth prevalence of 7/1,000,000 for the 300 cases reported so far. It is caused by mutations of the *EVCI* and *EVC2* genes and shows autosomal recessive inheritance with parental consanguinity in about 30% of cases (1).

The EVC phenotype is variable and affects multiple organs. Bone age is usually retarded (2). Skeletal dysplasia

is present at birth, being characterized by short limbs, especially the middle and distal segments, accompanied by postaxial polydactyly of the hands and sometimes the feet. Nearly all patients show severe dystrophy of the fingernails, which are markedly hypoplastic, thin, and often spoon-shaped (1). Congenital heart malformations are present in about 50-60% of cases (1,3,4). Some patients show mental retardation, even though this is not an integral component of the syndrome (1,5). The differential diagnosis includes asphyxiating thoracic dystrophy, achondroplasia, chondrodysplasia punctata, Morquio syndrome, and cartilage-hair hypoplasia, which can be differentiated on the basis of radiographic and clinical features (1). The disorder has been diagnosed prenatally using ultrasound and fetoscopy (1).

EVC shows a wide spectrum of oral manifestations, including malocclusion, labiogingival adhesions, labiogingival frenulum hypertrophy, accessory labiogingival frenula, a serrated appearance of the gingiva, dental transposition, diastema, conical teeth, enamel hypoplasia and hypodontia. Teeth may show premature eruption at birth, or premature exfoliation (1). Supernumerary teeth may also be present (2,6).

Case Report

A 5-year-old talkative girl with a chief complaint of pain in the left lower quadrant was referred to the Department of Pediatric Dentistry, Tabriz University of Medical Sciences.

The patient had been born to a mother who had suffered two miscarriages, 5 and 11 years previously, and had no siblings. Her parents were second cousins (Fig. 1) and in good general health.

Her stature was 86 cm, which was relatively short for age (Fig. 2a). The extremities were plump, and shortness

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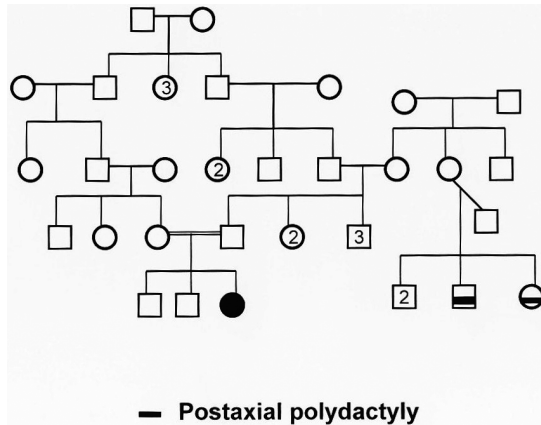


Fig. 1 Family pedigree.

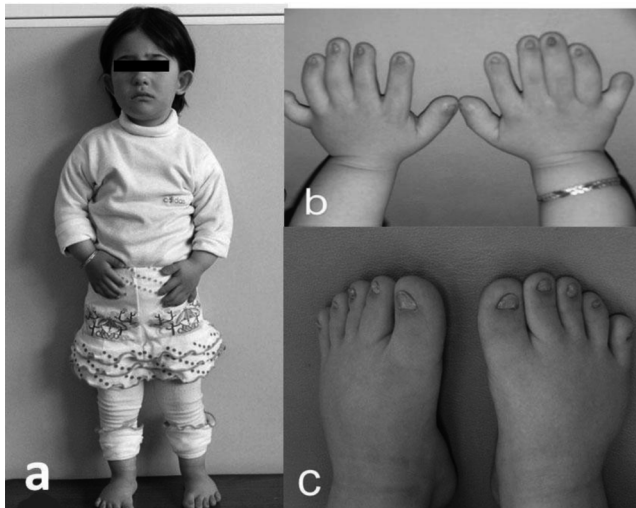


Fig. 2 (a) Short stature of the patient. The ratio of the trunk relative to the limbs is increased. (b) Hexadactyly and hypoplastic nails of the hands. (c) Hypoplastic nails of the feet.

of the limbs was evident. The hands and feet were wide and markedly deformed, with sausage-shaped fingers and dysplastic fingernails (Fig. 2b, c). Bimanual hexadactyly was noted on the ulnar side. The hair was fine and straight, but not sparse. The multiple clinical features of the patient revealed during examination by the dental team were strongly suggestive of a syndromic condition, leading us to suspect EVC syndrome. Therefore, the patient was referred to a pediatric hospital for further examinations.

A comprehensive examination was completed by a team of multi-disciplinary pediatric specialists. Medical examinations using echocardiography and ultrasonography revealed no evidence of systemic involvement such as congenital heart malformation or disorders of the lungs,

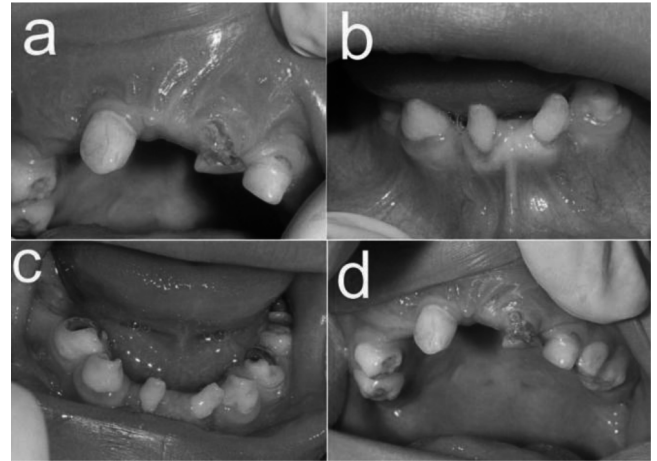


Fig. 3 (a) Fusion of the middle portion of the upper lip to the maxillary gingival margin. (b) Accessory labi gingival frenula and alveolar notches. Conical hypoplastic teeth with multiple caries in the lower (c) and upper (d) jaws. Note hypodontia in both arches.

kidneys, liver, pancreas and central nervous system. The patient's intellectual ability was within the normal range.

Intra-oral clinical examination revealed fusion of the middle portion of the upper lip to the maxillary gingival mucosal margin with absence of the mucobuccal fold, multiple accessory labi gingival frenula, conical anterior teeth, oligodontia of the primary dentition (absence of the lower right lateral incisor, lower left central incisor, upper left central incisor, and upper right lateral incisor and canine), multiple small alveolar notches on the crest of a thin alveolar ridge, and a serrated appearance of the gingiva (Fig. 3). The patient's parents reported no previous history of tooth extraction or natal tooth. An abnormal crown form, enamel hypoplasia and multiple caries were noted.

Panoramic radiography showed oligodontia of the permanent teeth. The tooth germs of the incisors, canines, the second premolar of the mandible and the maxillary right lateral incisor were absent (Fig. 4a). There was a supernumerary tooth in the left upper quadrant (Fig. 4b). The pulp chambers of the primary teeth had an increased apicoocclusal height and lacked the constriction at the level of the CEJ, resulting in a rectangular shape, and thus resembling the features of taurodontism (Fig. 4a).

Based on the clinical and radiographic findings of the dental and subsequent medical examinations, the medical team diagnosed the patient as having EVC syndrome. Written informed consent was obtained from the patient's parents to report the case.

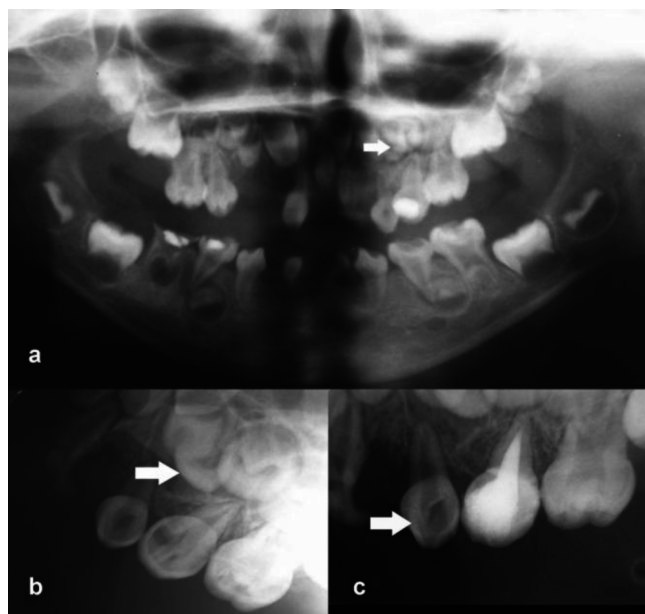


Fig. 4 (a) Panoramic radiograph showing primary molars with taurodontism. Hypodontia of the permanent dentition is evident. A supernumerary tooth (white arrow) is seen in the upper left quadrant on the panoramic and (b) periapical radiographs. (c) A large pulp chamber is evident in the primary upper left canine.

Discussion

The patient, living in a rural area, had not undergone any comprehensive examinations before being referred to the Department of Pediatric Dentistry, where oral findings in addition to other clinical features led us to suspect EVC syndrome. The present patient showed all of the typical clinical and radiographic oral features of EVC. Fusion of the upper lip with the gingival margin, characteristic of this syndrome, could be due to retardation of bone development (7) in the alveolar ridge, as is known to occur with other bones.

Multiple small alveolar notches on the crest of the thin alveolar ridge were also evident, involving the areas normally occupied by the lateral incisors (6,8). Each submucous cleft was marked by a moderately sized fibrous band, whose fibers appeared to incise the underlying alveolar process and extend across the mucobuccal fold into the lip. Between the two incomplete clefts and the moderately sized fibrous bands were numerous smaller fibrous bands which, when considered together, formed a continuous frenulum that reduced the extent of the attached gingiva and the labial sulcus. According to Gorlin et al. (1), this notching is a continuation of the normal serrated condition, which is present from the third to the seventh

month in utero.

In this patient, five primary and nine permanent teeth, other than the third molars, were absent. Such severe hypodontia beyond the incisor region has been reported in one case (6). Despite the reports of supernumerary teeth in EVC syndrome (6,9,10), the posterior supernumerary tooth in this case was a new finding not reported previously. It is interesting to note that, as seen on the panoramic radiograph, permanent teeth corresponding to the missing primary teeth were present. However, differentiation between supernumerary and actual permanent teeth was equivocal.

In patients with EVC syndrome, teeth that do erupt tend to be small, conically crowned, or resembling bicuspid with an accentuated cusp height and deep steep-sided fissure patterns. This may explain the high incidence of caries in affected patients (6), although enamel hypoplasia might be another reason. Taurodontism, a feature that has been reported previously in permanent and some primary molars (6,11,12), was found in all of the primary teeth in the present patient.

EVC syndrome requires multidisciplinary treatment planning to achieve satisfactory functional and esthetic results. The talkativeness of our patient, and another reported previously (6), may be a behavioral characteristic of EVC syndrome, and perhaps an important issue to consider for behavior management.

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