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Oral abnormalities in the Ellis-van Creveld syndrome: case report

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Abstract

Ellis-van Creveld syndrome, also called chondroectodermal dysplasia, is an autosomal recessive disorder characterized by disproportionate dwarfism, narrow thorax, polydactyly, and oral and dental abnormalities (Rimoin 1975; Simon and Young 1986). Congenital cardiac disease, usually a septal defect, and nail dystrophies are common. Anomalies of the central nervous system and urogenital tract also have been reported (Blackburn and Belliveau 1971; Rosemberg et al. 1983).

This syndrome is of interest to the pediatric dentist in that abnormal frenum attachments, hypodontia, malformed teeth, and malocclusion are invariably present (Biggerstaff and Mazaheri 1968). Natal and neonatal teeth, delayed eruption of permanent teeth, notching of the alveolar ridge, and a high caries rate are common (Gorlin et al. 1976; Smith 1982).

Literature Review

In 1940 Ellis and van Creveld described 3 cases of a disorder they called chondroectodermal dysplasia. Features reported included "chondrodysplasia" manifesting as acromelic dwarfism, polydactyly, cardiac abnormalities, and ectodermal dysplasia. Features of the ectodermal dysplasia included small dysplastic nails, fine scanty hair in two cases, and orodental abnormalities. One of the 3 patients had 2 natal teeth and the other 2 had delayed eruption of primary teeth. All patients exhibited hypodontia and conically shaped teeth. Obliteration of the maxillary anterior vestibule due to an abnormally wide labial frenum was described in 2 cases.

By 1981 a large number of cases, including more than 50 in the Amish of Lancaster, Pennsylvania, were reported. Many authors included descriptions of oral and dental abnormalities. Congenital heart disease, usually an atrial septal defect, was found to be present in as many as 60% of cases (McKusick et al. 1964). For this reason the term chondroectodermal dysplasia was replaced by Ellis-van Creveld (EvC) syndrome as the preferred designation. Cardiac and respiratory problems due to thoracic malformations were the major cause of death with more than one-third of patients dying in infancy. Many patients, however, live a normal or near normal life span.

Constant features of the EvC syndrome were polydactyly of the hands (and occasionally the feet); short-limbed, disproportionate dwarfism; and abnormal labial frenum attachments. Nails were usually small and dystrophic and urogenital anomalies were found in some patients. Intelligence of patients was normal and hair was not affected. Inheritance was found to be autosomal recessive (McKusick et al. 1964; McKusick 1973).

The most constant oral finding in the EvC syndrome is a broad labial frenum obliterating the maxillary labial vestibule. The mandibular labial vestibule often shows abnormal frenum attachments as well (Mitchell and Wadell 1958; McKusick et al. 1964; McKusick 1973; Eidelman and Rosenzweig 1965; Winter and Geddes 1967; Biggerstaff and Mazaheri 1968; Tzukert and Garfunkel 1978; Sarnat et al. 1980; da Silva et al. 1980). Hypodontia (usually involving maxillary and mandibular incisors), abnormally formed teeth (including conical incisors), and malocclusion were also constant findings (Mitchell and Wadell 1958; McKusick et al. 1964; McKusick 1973; Eidelman and Rosenzweig 1965; Winter and Geddes 1967; Biggerstaff and Mazaheri 1968; Sidhu and Subherwal 1968; Tzukert and Garfunkel 1978; Sarnat et al. 1980; da Silva et al. 1980). Natal and neonatal teeth, often lost spontaneously, were reported in many cases (McKusick et al. 1964; McKusick 1973). Delayed eruption was noted in 2 siblings (Mitchell and Wadell 1958), while enamel defects and high caries rate were reported in a number of cases (Mitchell and Wadell 1958; Winter and Geddes 1967; Biggerstaff and Mazaheri 1968; Sarnat et al. 1980; da Silva et al. 1980). Bilateral partial clefts or notching of the alveolar bone in the maxillary and/or mandibular lateral incisor region were found in 9 patients for whom radiographs

were available (Eidelman and Rosenzweig 1965; Winter and Geddes 1967; Biggerstaff and Mazaheri 1968; Tzukert and Garfunkel 1978; Sarnat et al. 1980). Other abnormalities described included 2 cases of double teeth (McKusick et al. 1964; McKusick 1973; Sarnat et al. 1980), 2 cases of mesiodens or supernumerary maxillary incisor (Winter and Geddes 1967; Sarnat et al. 1980), 2 cases of pulp stones (Winter and Geddes 1967; Tzukert and Garfunkel 1978), and 1 patient with taurodontism and premolar enamel invagination (Winter and Geddes 1967).

Case Report

Medical History

AE, a 2-year, 4-month-old white male was first seen at the University of Chicago Walter G. Zoller Memorial Dental Clinic at age 1 year, 3 months and again at 2 years, 2 months for radiographic examination of the dentition. The child returned for comprehensive dental examination with the chief complaint of a brown tooth that was difficult to clean.

The patient was the youngest of 3 children with a normal 11-year-old brother and 4-year-old adopted sister. Another sister with EvC syndrome died at age 6 months of congestive heart failure secondary to a 2-chambered heart. The family history was otherwise negative for the EvC syndrome.

The patient was the 7-pound, 5-ounce product of a term delivery following a difficult pregnancy. Diagnosis of the EvC syndrome was made by fetal ultrasound 4 months in utero. The child was noted to have dwarfed limbs and polydactyly (6 fingers on each hand). No cardiac, CNS, or genitourinary tract abnormalities were noted.

The medical history was positive for recurrent otitis media in infancy which resulted in placement of bilateral tympanostomy tubes under general anesthesia at age 8 months. AE was discharged, but returned to the hospital 2 weeks postoperatively for treatment of respiratory distress. He was treated and released with discharge medications of isoproterenol and theophylline as needed. Echocardiography at this admission confirmed a normal 4-chambered heart.

Later episodes of respiratory difficulties, probably due to small thoracic circumference, were treated on an outpatient basis. All medications were discontinued at age 19 months. At age 27 months supernumerary fingers were removed under general anesthesia without operative or respiratory complications.

Examination

AE was a short (80 cm), normocephalic, 2-year, 4-month-old white male, with short limbs, dysplastic finger and toe nails, normal facies, fine-textured blond hair and blue eyes (Fig 1).

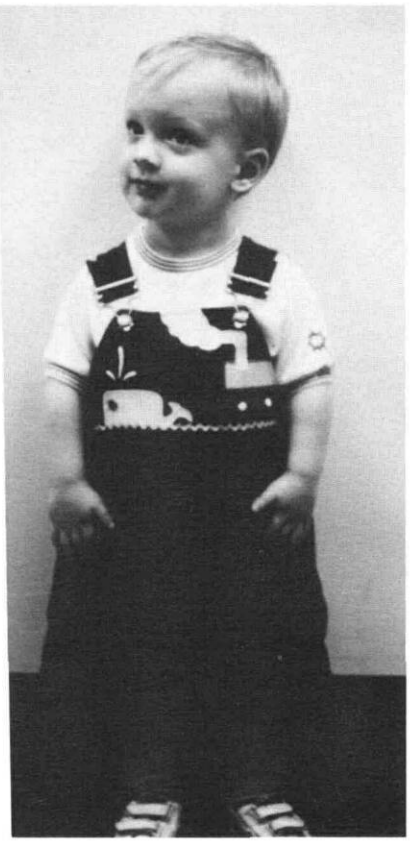


FIG 1. AE at age 3 years, 1 month.

Soft tissue examination revealed a broad maxillary labial frenum extending from canine to canine and largely obliterating the maxillary vestibule. Multiple, small, mandibular labial frenula were noted, but the vestibule seemed to be of normal depth. Seventeen of 20 primary teeth were present. All 8 primary molars were abnormal in morphology and incipient occlusal caries was present in the grooves of all maxillary molars (Fig 2, next page). Only 2 mandibular incisors were present and both were conical in shape. The maxillary primary right lateral incisor was missing. By the mother's report, this tooth erupted around 1 1/2 months of age and fell out shortly thereafter. The maxillary primary left lateral incisor was grossly carious and brown

AE's mother brushed his teeth daily, but continued to put him to bed with a bottle of milk or juice at night. Nursing bottle caries syndrome was discussed and the mother agreed to eliminate use of the nursing bottle. Appropriate oral hygiene measures were demonstrated and instituted. The patient was placed on a daily regimen of .04% stannous fluoride gel and the mother was instructed in its use. Due to the patient's age, presence of multiple caries, and poor behavior, treatment under general anesthesia was considered. The patient's pediatrician was consulted and the procedure was scheduled.

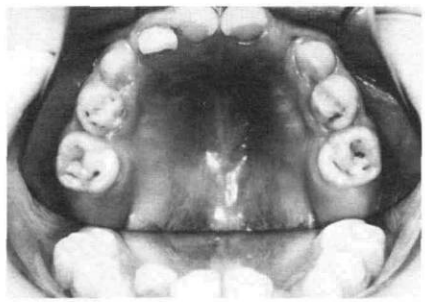
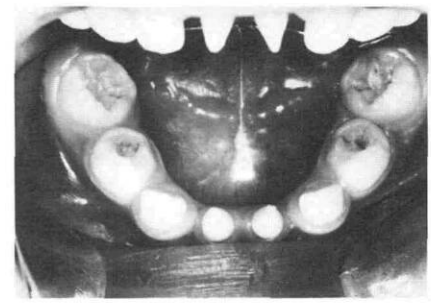


FIG 2a *(left).* Multiple abnormalities were present in the maxillary arch.

FIG 2b *(right).* Multiple abnormalities also were present in the mandibular arch.



Treatment

AE was admitted to the general pediatrics service for preoperative history, physical examination, laboratory work-up, and dental rehabilitation under general anesthesia the following day. All physical and laboratory findings were within the patient's normal limits.

Maxillary and mandibular periapical radiographs taken at age 15 months and a maxillary periapical radiograph from age 2 years, 2 months showed a mesiodens forming between the permanent maxillary central incisor tooth buds (Fig 3). A radiopaque structure was present in the right maxillary primary lateral incisor region.

Occlusion was examined and contacts were found to be mainly on the primary second molars (Fig 4). Seven periapical and 2 bite-wing radiographs were taken (Fig 5, next page), revealing the absence of tooth buds for the maxillary lateral incisors and all mandibular incisors. A small mesiodens was present between the permanent maxillary central incisor tooth buds, but the radiopaque structure in the maxillary right lateral incisor was absent and was assumed to have resorbed. Notching of the alveolar bone was present mesial to all 4 primary canines.

The throat was packed, the teeth were cleansed and examined, and additional carious lesions were noted on the occlusal of the mandibular first molars. Rubber dam isolation was used for all restorative procedures.

The left maxillary lateral incisor was restored using an acid-etch, light-cured, composite crown technique. The mandibular first molars had relatively normal occlusal anatomy and carious lesions were restored with amalgam.

The occlusal anatomy of the maxillary molars and mandibular second molars was grossly abnormal and included many fine accessory grooves (Fig 3). Use of amalgam restorative material would have resulted in a high risk of recurrent caries in unrestored grooves. Instead, an acid-etch, light-cure composite technique with dentin bonding agent and occlusal sealant was used.

Following a fluoride treatment and removal of the throat packs, the patient was extubated and brought to the recovery room where he continued to recover without complications. The mother was given postoperative instructions and the patient was discharged the following morning in apparent good health.

The mother failed to keep a 2-week postoperative visit due to the long distance from her home to the hospital, but reported that AE was doing well. Home care instructions were reinforced over the telephone.

At a recall examination 6 months later, no new carious lesions were found, all restorations were intact, and the mother was continuing a good home care program.

Discussion

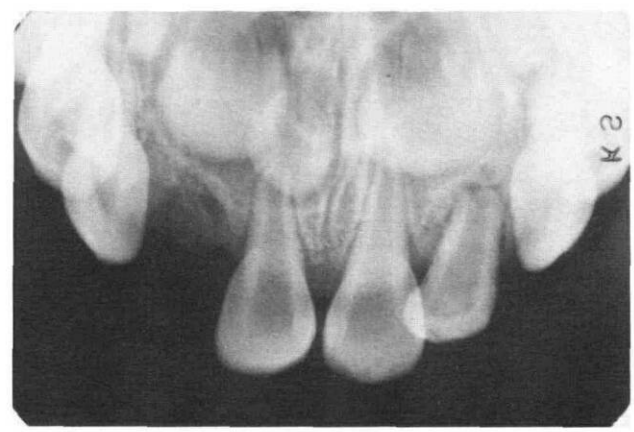


FIG 3. Maxillary periapical radiograph at age 2 years, 2 months reveals a developing mesiodens and radiopaque structure in the right lateral incisor region.

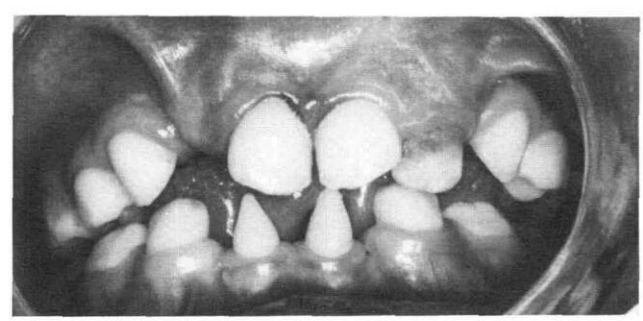


FIG 4. Occlusal contacts were mainly on the primary second molars.

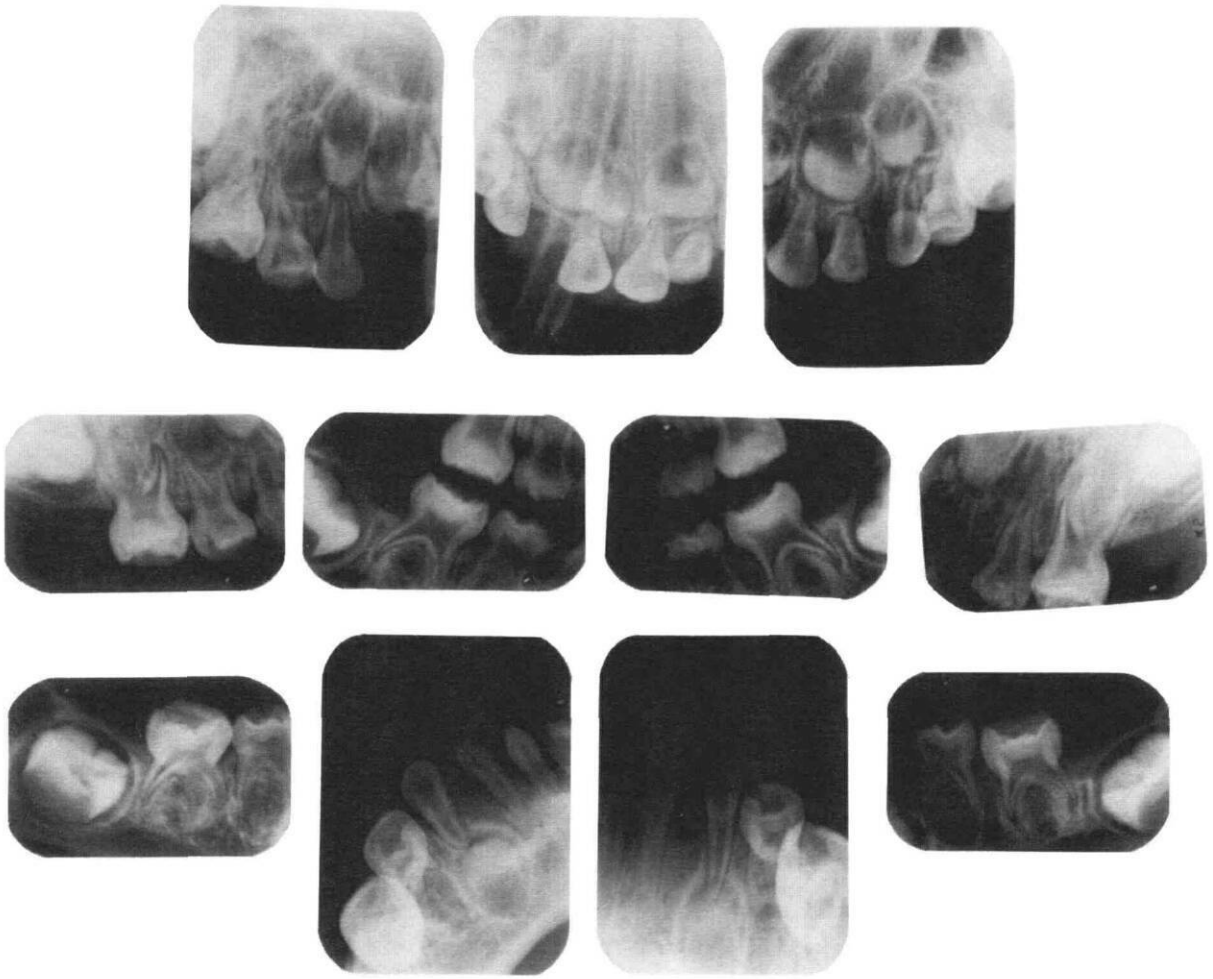


FIG 5. Radiographs obtained at age 2 years, 6 months revealed multiple oral and dental anomalies.

The patient in this report demonstrates many of the typical oral and dental features of the EvC syndrome. The broad maxillary labial frenum variously described as "partial harelip", "multiple small frenula", and "obliteration of the maxillary vestibule" is an almost constant finding, as are conical incisors, abnormally shaped molars, and malocclusion. Absence of some primary incisors, all permanent mandibular incisors, and permanent lateral incisors is also common. The abnormal occlusal anatomy of the molars with many pits and fissures, was partially responsible for the high caries rate, but use of the nursing bottle as a pacifier was a contributing factor, especially in the maxillary anterior region.

A neonatal tooth (maxillary right lateral incisor) was present, but as described in other cases (Ellis and van Creveld 1940; McKusick et al. 1964) was lost spontaneously. A radiopaque structure was present in this region between the ages of 15 and 24 months, but had disappeared by age 30 months. This structure probably represented the apical remnant of the lost neonatal tooth as described by Ooshima et al. (1986). This apical remnant probably was resorbed.

A less common finding in the EvC syndrome is the presence of a mesiodens. It has been reported only by Sarnat et al. (1980). The decision was made to preserve this supernumerary tooth due to its proximity to the permanent central incisor tooth buds and possible future need for extra teeth.

Conservative restorations were selected wherever possible due to the large pulp chambers of the primary teeth. Sealants and composite/sealant combinations should help prevent recurrent caries in the abnormal grooves. Effective home oral hygiene and a daily .04% stannous fluoride regimen controlled by the mother should also minimize future decay.

This patient's personal and family history highlight the need for a thorough cardiac and respiratory evaluation of all EvC syndrome patients. Cardiorespiratory problems are an important prognostic indicator and

also may require modification of dental treatment plans (e.g., SBE prophylaxis).

Future dental care for this patient will consist of a strong preventive program and restoration of caries as necessary. Long-term dental treatment probably will include a combination of orthodontics to align the permanent teeth and fixed bridgework (either conventional or resin bonded) to replace missing incisors. Crowns or composite build-ups may be necessary to modify abnormally shaped teeth.

At the time of the study Dr. Himelhoch was a clinical assistant professor, Zoller Dental Clinic and pediatrics, University of Chicago Medical Center; Dr. Mostofi is an associate professor and section head, oral pathology and radiology, University of Chicago Medical Center. Reprint requests should be sent to: Dr. Deborah A. Himelhoch, 11 Conrad Rd., Framingham, MA 01701.

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Frozen transplants

A Danish doctor has developed a method for transplanting teeth within a patient's mouth and from donors to recipients by freezing teeth and using them later.

The donors are usually young people having teeth pulled for orthodontic reasons. The teeth are sorted and classified according to the donor's tissue type as deternined by a blood test and then stored at -200° C.

Dr. Ole Schwartz of the Copenhagen Dental College moves teeth from the upper to the lower jaw, or from the back to the front of the mouth. He claims this technique could be most useful for people with cleft palates whose maxillary teeth are usually pulled prior to rebuilding the jaw. Now, the pulled teeth can be frozen and reinserted into the restructured jaw.