DUDAK SİNEŞİSİ VE POLİDAKTİLİSİ OLAN ÇOCUK OLGU: ELLIS-VAN CREVELD SENDROMU

A CHILD WTIH LIP SYNECHIA AND POLYDACTYLY: ELLIS-VAN CREVELD SYNDROME

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ÖZET

Ellis-van Creveld sendromu iskelet sistemi anomalileri ile karakterize herediter bir tablodur. Kondrodisplazi, ektodermal displazi, konjenital kardiak defektler ve polidaktili karakteristik özelliklerindendir. Alt alveoler arkta çentiklenme, üst dudak ve gingivomukozal marjinde füzyon, sıklıkla neonatal diş varlığı, oligodonti ve anterior kısımdaki dişlerde konik şekil bulunması gibi oral mukozal ve dental değişikliklerin varlığı Ellis-van Creveld sendromu tanısını kuvvetlendirmektedir. 13 aylık kız çocuğu bilateral postaksiyal polidaktili ve üst dudak ve gingivomukozal bileşkede füzyon deformitelerinin rekonstrüksiyonu nedeniyle tarafımıza refere edildi. Detaylı muayenesinde kondroektodermal displazi ve kardiak anomali saptandı. Aile genetik inceleme yapılmasını kabul etmedi fakat hastanın klinik bulguları Ellis-van Creveld Sendromu tanısını güçlü bir şekilde destekliyordu.

Anahtar Kelimeler: Ellis-van Creveld sendromu postaksiyal polidaktili, maksiller jinjiva labiyal füzyon

ABSTRACT

Ellis-van Creveld syndrome is a hereditary table especially characterized by skleletal system anomalies. It is characterized by chondrodysplasia, ectodermal dysplasia, polydactyly and congenital cardiac defects. The presence of oral mucosal and dental alterations like notching of the lower alveolar process, fusion of the upper lip with gingival mucosal margin, occasional presence of neonatal teeth, oligodontia and conical shape of anterior teeth will confirm the diagnosis of Ellis-van Creveld syndrome. A 13-month-old girl infant with postaxial polydactyly and fusion of the upper lip with gingivomucosal margin referred to our clinic for reconstruction of the deformities. Her detailed examination revealed chondroectodermal dysplasia and cardiac anomaly as well. The family refused a genetical investigation but the findings were strongly suggesting the diagnosis of Ellis-van Creveld syndrome.

Keywords: Ellis-van Creveld syndrome, postaxial polydactyly, maxillary gingivolabial fusion

INTRODUCTION

Chondroectodermal dysplasia is a rare mesenchymal – ectodermal dysplasia first described in 1940 by Richard W.B. Ellis and Simon van Creveld.[1] The exact prevalence of this illness is unknown. About 150 cases are described in the literature.[2] Ectodermal dysplasia is present up to 93% of them. Five different mutations have been associated with this syndrome.[3] Polydactyly of the feet is present in only 10% of the patients. A wide space is often present between the hallux and other toes.[4] The extremities are often plump. Acromelic and mesomelic shortness of limbs is often encountered. Shortening is most common in distal aspect of the limbs. Frequently the patient cannot make a tight fist.[5] 50 or 60% of the patients deal with congenital cardiac defects. Endocardial cushion defects and wide atrial septal defect like single atrium are the most common cardiac anomalies.[6] Genu valga .[7,8] curvature of humerus, talipes equinovarus,[9] talipes calcaneovalgus and pectus carinatum[10,5] with a long narrow chest with respiratory difficulties are the other features.[1]

Fusion of the middle portion of the upper lip to the maxillary gingival margin eliminating the maxillary labial vestibule or the presence of numerous frenula tethering the upper lip to gingiva are usually seen.[1] Hypodontia involving the maxillary and mandibular incisor region is a consistent finding, though supernumerary teeth have been reported in some cases.[11-13] Although most patients have normal intelligence, occasional central nervous system anomalies or mental retardation have been reported.[1]

CASE REPORT:

A 13- month-old girl referred to our clinic for reconstruction of polydactyly and lip fusion at the gingivobuccal sulcus. She was diagnosed with polydactyly at the 4th gestational month. The mother had a nonspeci-

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fic pregnancy and the parents were not relatives. A conventional birth at the 39th gestational week has been performed with ceaserian section. She weighed about 3,500 grams at birth. The family was offered a genetic counseling but they refused it after her birth.

Her gross examination on arrival revealed defective and deformed nails with bilateral postaxial ulnar polydactyly on hands, fusion of the upper lip to the maxillary gingival margin and notching of the lower alveolar process with no congenital dentitions. She weighed 13 kgs and she was 78 centimetres tall. She was not mentally different from her age group. There was neither cyanosis nor clubbing. The nails of both hands and feet demonstrated an extreme degree of dysplasia. Her hair was fine and sparse, her scalp appeared normal.

There was no tooth eruption in the upper jaw. Sulcus formation between maxillary gingiva and upper lip was absent (Figure 1). The labial mucosal membrane was attached along the maxillary mucosa where the incisor and canine teeth normally arise from. Lower jaw was also defective of teeth eruption. The level of the gum pad was interrupted by a depression on each side of the lateral incisor region.

The skull and face appeared clinically and radiologically normal and did not demonstrate the characteristics typically seen in achondroplasia.

There was a disproportinate dwarfism in the upper and lower extremities as seen in the achondroplasies (Figure 2). There were six digits on each hand (ulnar polydactyly). The terminal phalanges of the fingers were poorly developed. A sixth metacarpal bone was present on the right side and there was fifth and sixth metacarpal bone fusion on the left side (Figure 3).

Her echocardiography demonstrated patent foramen ovale. She had no complaints related to this anomaly. The parents were offered for genetic counseling once again but because of cultural reasons they refused it.

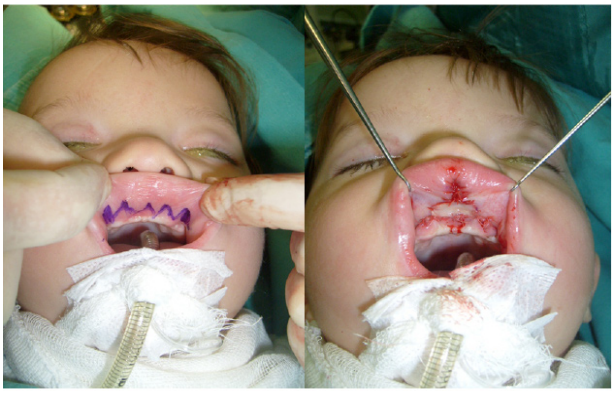

Figure 1. Lip synechia of the patient.

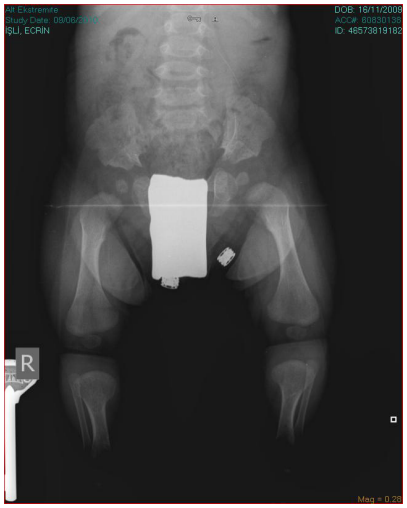

Figure 2. Radiographic view of the lower extremities.

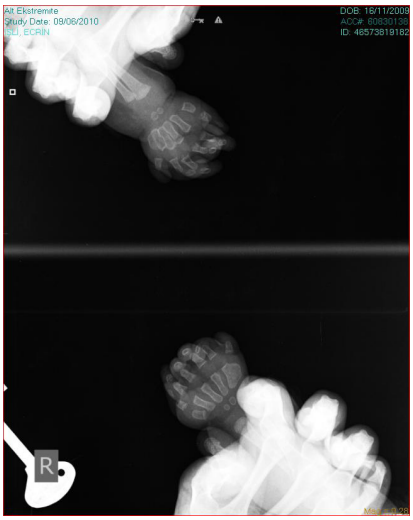

Figure 3. Preoperative radiograph of the right (lower view) and left (upper view) hands.

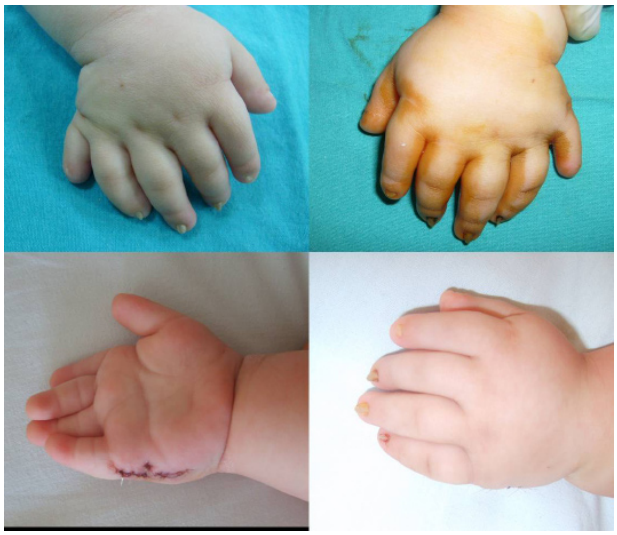

Figure 4. Upper views; preoperative pictures of the hands, Lower views; postoperative pictures of the hands.

After birth, the cardinal features usually present are: 1) disproportionate small stature with increasing seve-

rity from the proximal to distal portions of the limbs, and shortening of the middle and distal phalanges (Figure 3-4); 2) polydactyly affecting hands (uni [exceptional[14]] – or, usual, bilateral) (Figure 3-4) and, occasionally, the feet; 3) hidrotic ectodermal dysplasia mainly affecting the nails, hair and teeth (Figure 1); 4) congenital heart malformations occurring in about 50–60% of the cases and comprising of single atrium, defects of the mitral and tricuspid valves, patent ductus, ventricular septal defect, atrial septal defect and hypoplastic left heart syndrome. The presence of congenital heart disease may support the diagnosis of the EVC syndrome and appears to be the main determinant of longevity.[15,16] Four of the cardinal features were found in our patient; shortening of the middle and distal phalanges, bilateral polydactyly, dysplastic nails, hair and teeth and mildly congenital cardiac defect.

Surgical technique

The patient was examined with preoperative tests and there was no contraindication for the operation. In the operation theatre the right sixth finger was desarticulated from the level of the carpometacarpal joint. The left sixth finger was desarticulated from the level of the metacarpophalangeal joint. The ulnar side of the fusioned metacarp was osteotomisied and the ulnar contour of the hand was corrected. The extansor tendons and abductor digiti quinti mucle were sutured to the fifth finger on suitable anatomic location bilaterally. Excessive skin was excised and made single z plasty on each side of the incision. Care was taken not to bring the hypothenar skin too dorsally. The hands were splinted postoperatively (Figure 4).

The upper lip mucosa was marked for w plasty to create a sulcus and elongate the buccal mucosa of the lip. After the flaps were incised, the sulcus was deepened and the resulting defect was reconstructed with a mucosal greft harvested from the lateral buccal region. The donor site was primarily sutured. The patient was discharged from the hospital two days after the surgery without any complications.

DISCUSSION

Ellis-van Creveld syndrome, also called chondroectodermal dysplasia, is a rare occurence inherited as an autosomal recessive disease.[6] Family-based genetic studies identified human mutations in previously unknown genes, EVC and LBN (EVC2), which are located head-to-head on chromosome 4p16.2.[17] The majority of the cases were characteristically seen in two particular inbred population from the Amish community of Lancaster County, Pennsylvania, U.S.A. and Kerala region, India.[1,4] Five different mutations have been associated with this syndrome.[3] The disease is usually well defined at birth, besides it can be diagnosed at the prenatal period.[18] Nearly half of the cases dies at the newborn period.[4]

The disease is evaluated in Short Rib Polydactyly Syndromes (SRPS) and characterized with chondrodysplasia, ectodermal dysplasia, acromelic and mesomelic extremity shortness, postaxial polydactyly, small chest, long trunk and congenital cardiac defects.[6] The most important skleletal findings are acromelic dwarfizm and polydactyly. The patients reaching adulthood may be up to 115-150 centimeters tall. Polydactyly is seen nearly in all cases at the upper extremity, however less than 10 % at the lower extremity.[4]

Fusion of the middle portion of the upper lip to the maxillary gingival margin eliminating the maxillary labial vestibule or the presence of numerous frenula tethering the upper lip to gingiva are other features of the syndrome.[1]

Hypodontia involving the maxillary and mandibular incisor region is a consistent finding though supernumerary teeth have been reported in some cases.[11-13] Patients can be diagnosed in any age with odontogenic and malocclusion problems wich interests orthodontists and maxillofacial surgeons.

Fifty to sixty percent of the patients demonstrate congenital cardiac defects. Endocardial cushion defects and wide atrial septal defects, like single atrium, are the most common cardiac anomalies.[6] However, additional clinical findings affecting other organs (lungs, kidneys, liver, pancreas and central nervous system) may occasionally be observed.[2,19,20] although these were not diagnosed in our case. Genitourinary anomalies such as renal agenesis and dysplasia, ureterectasia and nephrocalcinosis are usually present in 20% of the cases.[21] Exceptionally, hematological anomalies have been reported. In the literature, one case with dyserythropoiesis and another associated with perinatal myeloblastic leukemia are described.[2,22]

Although most patients have normal intelligence, occasional central nervous system anomalies or mental retardation have been reported.[1]

In infancy and early adulthood, general and specialized pediatric follow-up studiesare also required: the short stature is considered resulting from chondrodysplasia of the legs and the possible treatment with growth hormone is considered ineffective. It is important to notice, however, that the association of growth hormone deficiency and EVC has been reported in one patient and, in this case, the growth hormone treatment had a favorable effect on growth.[23] The possibility of bone deformity, especially knee valgus with depression of the lateral tibial plateau and dislocation of the patella,[24] needs regular orthopedic follow-up study. Dentists play an important role in control of dental and oral manifestations. Dental treatment must be performed under prophylactic antibiotic coverage with regard to the high incidence of cardiac defects in EVC

patients.

Nearly half of the Ellis-van Creveld Syndrome patients are lost in the newborn period because of cardiorespiratory problems. Most of them have normal intelligence. The maximal length is not more than 150 centimeters. The therapy usually involves corrective surgery for dental problems, polydactyly, genu valgum and congenital cardiac defects. Our patient demonstrated mild characteristics of the syndrome. She had corrective polydactyly operation and gingivobuccal lip dehiscence from the maxillary mucosa and sulcus formation. She was a lucky kid with no major cardiac problem. Her findings strongly suggested the syndrome but her genetic counseling could not be done. When polydactyly is associated with chondroplasia, gingival anomalies, congenital heart disease and ectodermal dysplasia, there should be a high suspicion of this syndrome and associated anomalies should be investigated.

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