

Ellis–van Creveld Syndrome with Developmental Delay

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Abstract

Ellis–van Creveld (EVC) syndrome also known as chondro-ectodermal dysplasia or meso-ectodermal dysplasia, is a rare autosomal recessive syndrome. EVC belongs to the short rib-polydactyly group. The genetic defect is located at chromosome 4p16. Two different mutations EVC1 and EVC2 have been identified. All embryonic layers appear to be involved in EVC syndrome. This syndrome is characterized by skeletal and ectodermal dysplasia. The principal features of this syndrome are chondroectodermal dysplasia, polydactyly and congenital heart defects. We report a 10-month-old female child with EVC syndrome with all the classical features and developmental delay, which has not yet been reported with this syndrome.

Key words: Common atrium, Developmental delay, Ellis–van Creveld syndrome, Oral anomalies

INTRODUCTION

Ellis–van Creveld (EVC) syndrome also known as chondro-ectodermal dysplasia or meso-ectodermal dysplasia was first described by Ellis of Edinburgh and Simon van Creveld of Amsterdam in 1940.¹ It is a rare autosomal recessive disease resulting from a genetic defect located at chromosome 4p16. Two different mutations EVC1 and EVC2 have been identified.²⁻⁴ This syndrome is most prevalent in the Amish population occurring in 1/5000 live births and the birth prevalence in non-Amish population is estimated to be 7/10,00,000.^{5,6} The principal features of this syndrome are chondroectodermal dysplasia, polydactyly, and congenital heart defects. The patients have a small stature, short limbs, fine sparse hair and hypoplastic fingernails. Oral manifestations include multiple musculo-fibrous frenula, dental transposition, conical teeth, hypoplasia of the enamel, hypodontia, and malocclusion. The teeth can erupt and exfoliate prematurely.^{2-4,7,8}

CASE REPORT

A 10-month-old female was the first child born of a non-consanguineous marriage presented with hyperdynamic precordium since birth, sweating on the forehead, decreased activity and delayed developmental milestones. Perinatal history was normal except she had natal teeth that were later shredded. Family history was normal.

On examination she had weight 6.5 kg, length of 62 cm, occipitofrontal circumference of 41 cm, chest circumference of 39 cm, upper segment/lower segment ratio of 1.57:1, long narrow dysplastic chest and abdomen (Figure 1), fusion of upper lip to underlying gums (Figure 2), abnormal upper and lower gums (Figure 3), postaxial polydactyly of both hands and hypoplastic nails (Figure 4), small pelvic bones, and genu valgum (Figure 5). A wide space is seen between the hallux and other toes (Figure 6). X-ray hip showed small pelvic bones. Chest X-ray showed pulmonary plethora (Figure 5). Electrocardiogram showed superiorly oriented QRS axis with left axis deviation (Figure 7). 2D-echocardiography showed single atrium. No anomalies in the analytic tests including routine blood and ultrasound abdomen were found. No genetic study had been made. All these findings confirm EVC syndrome. Patient was diagnosed as EVC syndrome with congenital heart disease.

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Figure 1: Long narrow dysplastic chest and abdomen

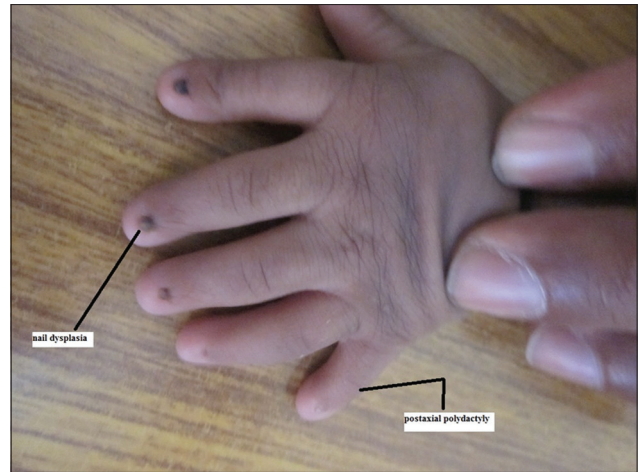


Figure 4: Child with Ellis-van Creveld syndrome showing postaxial polydactyly and nail dysplasia

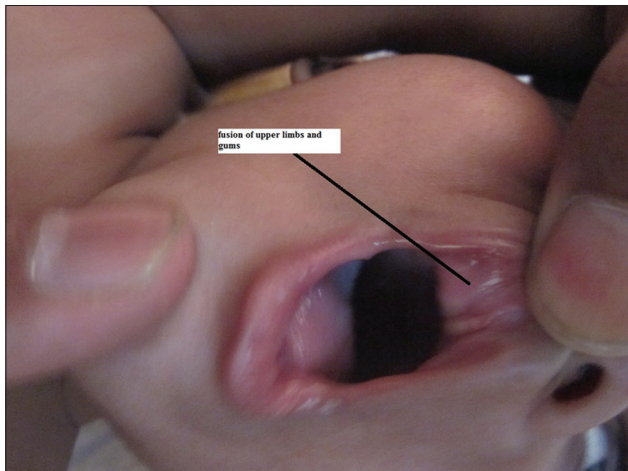


Figure 2: Child with Ellis-van Creveld syndrome showing fusion of upper lip to gums

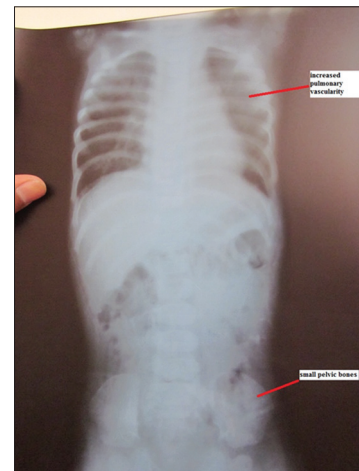


Figure 5: X-ray showing increased pulmonary vascularity and small pelvic bones

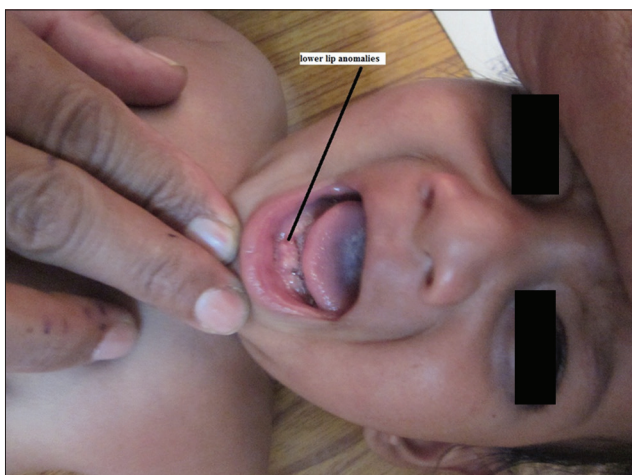


Figure 3: Child with Ellis-van Creveld syndrome showing lower lip anomalies

with congestive heart failure with development delay and was treated and discharged.

DISCUSSION

EVC syndrome also known as chondro-ectodermal dysplasia is a skeletal and an ectodermal dysplasia. All embryonic layers appear to be involved in EVC syndrome. The signs of ectodermal dysplasia are usually limited to nails, teeth and gums, although some cases with eye and neural involvement have been described. Abnormalities of the skeletal system and the heart and in some patients of the kidneys indicate the mesodermal involvement. Endodermal involvement is not very common.⁹ The syndrome can be diagnosed during the prenatal period, starting from the 18th week of gestation, by ultrasonography, or later by clinical examination after birth.

The skeletal dysplasia presents at birth with short limb, especially the middle and distal segments (acromelic and mesomelic), postaxial polydactyly, wide hands and feet, sausage-shaped fingers of hands and fingernail dysplasia



Figure 6: Wide gap between hallux and toes

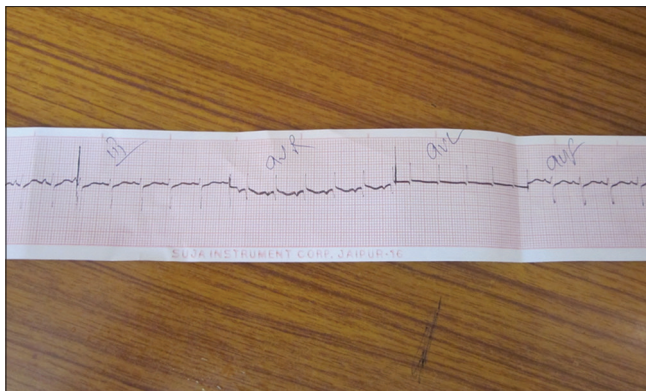


Figure 7: Electrocardiogram showing left axis deviation and superior QRS axis

are seen. The thorax is usually narrow with pectus excavatum, lumbar lordosis and genu valgum. A wide space is often seen between the hallux and other toes. The hair is sparse and fine. All the above mentioned skeletal features were present in our case. Adult height ranges from 109 cm to 152 cm.

Oral manifestations seen are dental anomalies including neonatal teeth, absent eruption, delayed eruption, premature loss of teeth, malocclusion, hypodontia, and appearance of conical teeth, hypoplasia of the enamel and multiple musculo-fibrous frenula and upper lip defects which matched our case.

Common congenital heart malformations include atrial septal defect, single atrium or common atrium and rarely ventricular septal defect, patent ductus arteriosus, hypoplasia of aorta. Our case had common atrium, which is seen in 40% of cases.

The cognitive and motor developments of patients affected by EVC syndrome are normal.^{2,4} However in our case, there was a significant developmental delay in all 4 aspects.

Skeletal radiograph reveal short tubular bones with clubbed ends, especially the proximal tibia and ulna. Carpal bones display extra ossification centers and fusion.

EVC belongs to the short rib-polydactyly group (SRP). These SRPs are all autosomal recessive disorders that have been classified into types (Saldino-Noonan syndrome, Type I; Majewski syndrome, Type II; Verma-Naumoff syndrome, Type III; Beemer-Langer syndrome, Type IV; and Jeune dysostosis). They are characterized by hypoplastic thorax due to short ribs, short limbs, frequent polydactyly and visceral abnormalities, and are discussed prenatally. Radiographically and histologically, SRP III (Verma-Naumoff syndrome) most resembles some forms of EVC.^{10,11}

About 30% patients die of cardiac or respiratory problems during infancy. The management of EVC is multidisciplinary. Symptomatic management is mostly required in the neonatal period, including treatment of the respiratory distress due both to the narrow chest and heart failure. Neonatal teeth should be removed because they may impair the feeding. In infancy and early adulthood, general and specialized pediatric follow-up are also required: The short stature is considered resulting of chondrodysplasia of the legs and the possible treatment with growth hormone is considered ineffective. The possibility of bones deformity, especially knee valgus and dislocation of the patella, needs regular orthopedic follow-up.¹²

CONCLUSION

EVC syndrome is a skeletal and an ectodermal dysplasia. Dentists play an important role in the control of dental and oral manifestations. Dental treatment must be performed under prophylactic antibiotic coverage with consideration for the high incidence of cardiac defects in EVC patients.

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