

Case Report

Chondroectodermal dysplasia (Ellis-van Creveld syndrome)

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ABSTRACT

Ellis-van Creveld syndrome (EVC) is a very rare mesenchymal-ectodermal dysplasia. This was first described in 1940 by Richard W. B. Ellis and Simon van Creveld. This rare condition is inherited as an autosomal recessive trait with variable expression. It is also known as mesoectodermal dysplasia or chondroectodermal dysplasia. The main features of this syndrome are short ribs, polydactyly, growth retardation, and ectodermal and heart defects. It is a rare disease with approximately 150 cases reported worldwide. It was found to be more common among the Amish. But sporadic cases have been reported from all over the world including India. The generalized dysplasia of endochondral ossification is because of a mutation in a novel gene on chromosome 4p16. Mutations of the EVC1 and EVC2 genes, located in head to head configuration on chromosome 4p16 have been identified as a causative factor

Keywords: Chondroectodermal dysplasia, EVC, Hypodontia

INTRODUCTION

The skeletal dysplasia presents at birth with short limbs especially the middle and distal segments accompanied by postaxial polydactyly of the hands and sometimes feet also. Nail dysplasia and dental anomalies constitute the ectodermal dysplasia. These are the natal tooth neonatal teeth absent and premature loss of teeth and upper lip defects.¹ Common manifestations also include atrial septal defects, ventricular septa defects and other congenital heart defects.^{2,3}

Skeletal radiographs reveal short tubular bones with clubbed ends especially proximal tibia and ulna. Carpal bones display extra ossification centers and fusion; cone shaped epiphysis is evident in hands.

This syndrome is more common among the Amish community but sporadic cases have been reported worldwide. Parental consanguinity is found in 30% of the cases. The prevalence outside Amish community is 7/10,00,000.⁴ Prenatal abnormalities like narrow thorax, shortening of long bones, hexadactyly and cardiac defects

can be detected by antenatal ultrasonography. After birth, cardinal features are short stature, short ribs, polydactyly, and dysplastic fingernails and teeth.

Congenital heart defects frequently associated are atrial septal defect and ventricular septal defect and occur in about 60% of cases. Cognitive and motor development is normal. This rare condition is inherited as an autosomal recessive trait with variable expression. Mutations of the EVC1 and EVC2 genes, located in a head to head configuration on chromosome 4p16, have been identified as causative factors in the etiology of this disease.

CASE REPORT

This patient 11-years-old male child reported to our outpatient department with abnormally shaped teeth (Figure 1) in the mouth along with missing lower front teeth. Further enquiry from the parents revealed that the teeth had never erupted. He was born with natal teeth which were extracted. History of consanguinity was positive with his parents being the first cousins. The pedigree history revealed that the patient's aunt and brother

were affected by the same condition indicating a recessive pattern running in the family. On examination the patient was malnourished, with very short stature measuring 112 cm (<3 SD). There were very sparse hairs on the scalp (Figure 1, 3 and 4). He also had dysmorphic facies (Figure 4) with narrow shoulders. His axial development was essentially normal. The distal extremities especially fingers and toes were small. polydactyly and dystrophic nails were not present. The child had a small ventricular septal defect.



Figure 1: Anodontia, hypodontia, sparse hair.



Figure 2: Peg like teeth, anodontia.



Figure 3: Thin sparse hair and skin dryness.



Figure 4: Dysmorphic features.

DISCUSSION

The usual clinical features of EVC consists of disproportionate dwarfism, bilateral postaxial polydactyly, ectodermal dysplasia with thin sparse hairs, small nails, thin sparse hair and hypodontia and abnormally formed teeth, congenital heart diseases, single atrium and ventricular septal defect.^{4,7}

It is genetic disorder with autosomal recessive transmission most often described in families with history of consanguinity. resulting in a child with the same condition.^{4,12} Prenatal diagnosis after 18 weeks of gestation can be done by ultrasonography and later by clinical examination after birth.^{4,13}

Additional clinical features include genu valgum. The intelligence is usually normal.^{4,9} Oral manifestations in syndrome are remarkable and constant. The most common finding is a fusion of the anterior portion of the upper lip to the maxillary gingival margin, so that no mucobuccal fold exists, causing the upper lip to present a slight V-notch in the middle as was in this case figures.^{3,5,8} The anterior portion of the lower alveolar ridge is often serrated and multiple small and conical molars have abnormal cusps or accessory groove.^{4,7} And sometimes hypoplastic enamel is seen. Congenitally missing primary and permanent teeth, dysmorphic conical-shaped roots and delay in eruption have also been reported.^{5,11} All these features were present in this case.

The management of EVC is multidisciplinary. Management during the neonatal period is mostly symptomatic involving. treatment of the respiratory distress due to narrow chest and heart failure. Orthopedic follow-up is required to manage the bony deformities. Professional dental care is a mandatory requirement for oral manifestations. Prognosis in these children is directly related to the respiratory difficulties in the first months of life due to thoracic narrowness and possible. It is very difficult about the final height in these children. Approximately 30% of patients die of cardiac or respiratory problems during infancy life span is otherwise normal. Adult height ranges from 119 cm- 161 cm.¹

CONCLUSION

Children suffering from EVC syndrome require a continuous and coordinated team approach for the management with a regular follow up of by pediatricians, surgeons, cardiologists, dentists, pedodontologist, pulmonologists, orthopedists, urologists, physical and occupational therapists and/or other health care professionals. In other words EVC syndrome requires multidisciplinary therapeutic planning. Genetic counseling is recommended for affected children and their parents. Prognosis is reasonably good as the cognition domain is unaffected and they can lead a normal or near normal and productive life.

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