CASE REPORT

Disproportionate Short Stature with Multisystem Involvement – Ellis-van Creveld Syndrome

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ABSTRACT

Ellis-van Creveld syndrome is a rare form of mesenchymal - ectodermal dysplasia. It is an autosomal recessive disorder characterized by disproportionate short stature, postaxial polydactyly, ectodermal dysplasia and congenital heart defect. This case report presents two cases with classical clinical findings along with some unusual features including rhizomelic limb shortening, global developmental delay and bilateral lens subluxation not reported previously.

Key words: *Skeletal dysplasia. Short stature. Cardiac defect. Ellis-van Creveld syndrome. Mesenchymal-ectodermal dysplasia. Lens subluxation.*

INTRODUCTION

Ellis-van Creveld syndrome is a rare form of mesenchymal-ectodermal dysplasia. It has an autosomal recessive inheritance caused by EVC gene located at 4p16. EVC 2 gene has also been described as head on head mutation at same location.[1] This is a rare condition with high family clusters reported in an old order Amish Community of Lancaster County, Pennsylvania, USA.[2]

Around 350 cases have been reported in literature presenting with features of chondroectodermal dysplasia.[2] The classical tetrad includes chondrodystrophy, polydactyly, ectodermal dysplasia and cardiac anomalies. Other clinical findings frequently reported with the syndrome include oral (absence of mucobuccal fold, short upper lip, serrated lower alveolar ridge), dental (neonatal teeth, partial anodontia, enamel defects, malocclusion) and genitourinary anomalies.[3] Uncommon findings reported in literature include cor triatrium, double orifice mitral valve, syndactyly, airway cysts, lung anomalies, strabismus, retinitis pigmentosa and dyserhythropoiesis.[4] Early deaths have been reported during infancy associated with respiratory problems mainly due to severe chest narrowness or congenital cardiac lesions.[2]

This report presents two cases of this rare dysplasia with some unusual features.

CASE REPORT

Case 1 was a 4-year-old boy who presented with decreased vision in both eyes for the last 3 months. He belonged to consanguineous parents, having intrauterine growth restriction in antenatal period and presently short for his age. No other siblings had any similar complaints. He was disproportionately short (72.4 cm with US/LS:1.48), having irregular dentition, abnormal shaped teeth, absent mucobuccal fold, hypoplastic dysplastic nails, narrow chest, bilateral postaxial polysyndactyly both hands, inability to form fist, genu valgum deformity, and hypospadias (Figures 1, 2). He also had a bulging pre-cordium with grade 3/6 pansystolic murmur at left sternal border.

Skeletal survey revealed rhizomelic limb shortening, bony ulnar polysyndactyly, fused carpel bones, small iliac crests and dome shaped upper end of tibia. Orthopantogram showed absent permanent dentition. Echocardiography showed complete atrioventricular septal defect with abnormal tricuspid valve. Ultrasonography revealed no structural abnormality of urinary tract. Eye evaluation revealed bilateral posteriosuperior lens subluxation. Developmental profile showed fine and motor age of 22 months, cognition of 28 months and language of 30 months using Portage developmental assessment materials.

Case 2 was an 8-year-old boy presenting with recurrent chest infections and failure to thrive since 3 years of age. He belonged to related parents with no adverse prenatal, natal or postnatal history. He was the only sibling with history of two neonatal deaths of unknown cause. He was a developmentally delayed, disproportionately short boy (102 cm with US/LS:1.57), with absent maxillary central incisors, serrated alveolar ridge, abnormal shaped teeth, absent mucobuccal fold, dystrophic nails, narrow chest, post axial polydactyly, genu valgus deformity and wide gap between 1st and 2nd toe (Figure 3). He had a bulging precordium with right ventricular heave and grade 3/6 ejection systolic murmur in the left upper sternal border.

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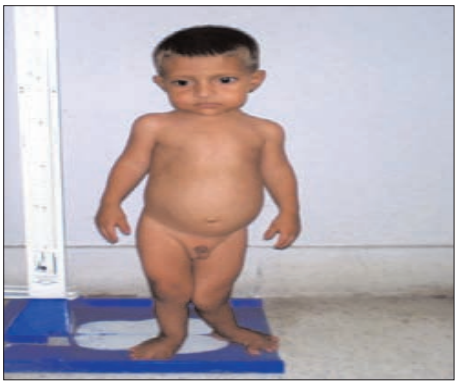

Figure 1: Case 1 having disproportionate short stature, rhizomelic limb shortening, narrow chest, genu valgus, nail hypoplasia, hypospadias and post axial bony polysyndactyly.

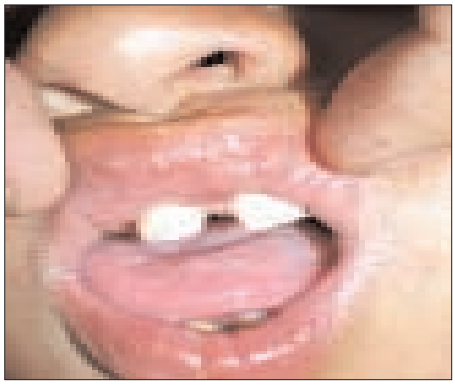

Figure 2: Case 1 showing absent mucobaccal fold.

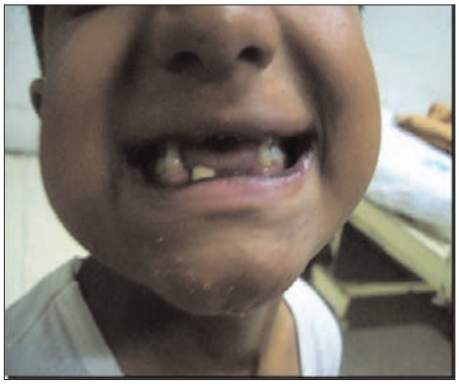

Figure 3: Case 2 showing serrated alveolar ridge and abnormal dentition.

Skeletal survey revealed rhizomelic limb shortening, funnel chest, bony ulnar polydactyly, small iliac crests and dome shaped upper end of tibia. Orthopentogram confirmed absence of maxillary incisors and malformed permanent dentition. Echocardiography revealed septum primum atrial septal defect with moderate pulmonary hypertension. Developmental profile found gross and fine motor skills at 65 months, cognition at 30 months and language at 34 months using Childhood Adaptive Behaviour Scale for Children.

DISCUSSION

The condition was first described by Richard Ellis and Simon van Creveld in 1940.[1] In the general population, the incidence is one in 60,000 live births and much lower in the Asian population. Only around 15 cases have been reported from the Indian subcontinent.[5] Parental consanguinity among the reported patients is as high as 70%. The reported cases had the classical tetrad with certain unusual features. The chondrodystrophy results in disproportionate short stature and mesomelic limb shortening. Both reported cases had rhizomelic and acromelic limb shortening sparing the forearms. Post-axial polydactyly was present in both children. Case 1 also had polysynmetacarpalism. Ectodermal dysplasia involving dysplastic nails and dental anomalies were present in both cases. Case 2 also had serrated upper alveolar ridge while previous reports showed mostly mandibular ridge serration. Cardiac anomalies in both cases were atrioventricular canal defect. Narrow chest and genu valgum deformity commonly observed in this syndrome were present in both cases. Genitourinary abnormalities are occasionally found including hypoplastic penis, renal agenesis and megaureter. Hypospadias found in Case 1 was also among rare findings described with the syndrome previously. Mental retardation documented in a few cases was secondary to CNS structural anomalies. Both cases had global delay in all fields of development despite the absence of any CNS anomaly on neuroimaging. Mild mental retardation has been documented in one pedigree with allelic deletions.[6] Significant developmental delay has not been described with this syndrome as well. Motor development comparatively more affected due to skeletal abnormalities. Case 1 had bilateral lens subluxation which is not previously reported with the syndrome.

Identification of the obvious dysmorphic features of this syndrome as early as possible is essential to prevent early deaths due to chest narrowness or congenital cardiac defects. Planning appropriate interventions for dental anomalies, cardiac defects and infective endocarditis prophylaxis is only possible with early identification. There has been a reported case of Ellis van Creveld syndrome where dental procedure was done without infective endocarditis prophylaxis as the cardiac lesion was never diagnosed.[2] In Case 2, the atrial septal defect was left unnoticed and the patient had started to develop pulmonary hypertension. Growth hormone therapy may improve height. Limited data does not allow any conclusions but the few patients studied have shown improved growth velocity with hormone therapy.[7]

Early detection of a constellation of features suggestive of Ellis-van Creveld syndrome is essential for establishing accurate diagnosis and early management as the condition involves multiple systems with little variation.

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