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Case Report

Pregnancy in Ellis Van Creveld syndrome women: a rare case report

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ABSTRACT

Ellis Van Creveld (EVC) syndrome also known as chondroectodermal dysplasia is a rare genetic disorder characterized by congenital heart defect, short ribs, polydactyly, dwarfism, deformed teeth and genu valgum. It is a rare disease with approximately 150 cases reported worldwide. The exact prevalence in India is not known, but the syndrome seems more common among Amish population of Pennsylvania in the United States of America (USA). Heart defects occur in about 60% of cases. Cognitive and motor development is normal. It shows autosomal recessive trait with variable expression. The patients with this syndrome rarely survive into adulthood but here we report a case of 25 years old pregnant lady with Ellis Van Creveld syndrome.

Keywords: Heart defects, Polydactyly, Dwarfism, Chondroectodermal dysplasia, Autosomal recessive

INTRODUCTION

In 1940, Richard W. B. Ellis of Edinburgh and Simon van Creveld of Amsterdam first described Ellis–van Creveld syndrome (EVC).[1] EVC is an autosomal recessive skeletal dysplasia, with inter- and intra-familial variability, characterized by short ribs, small chest, short limbs, disproportionate dwarfism, postaxial polydactyly, congenital heart malformation and dysplastic teeth and nails. Mutations in the EVC1 and EVC2 genes are associated with this syndrome. the genetic defect located in chromosome 4p16.[2] It is also known as chondroectodermal dysplasia. The syndrome is characterized by high mortality in early life, with 50% of deaths occurring during infancy.[3] It is a rare disease with approximately 150 cases reported worldwide. The exact prevalence in India is not known where as prevalence of the disease varies widely in general population of 1 in 60,000 live births in the United States of America (USA) to 1 in 150,000 live births in European countries, large number of cases were seen in Amish community of USA by McKuisk in 1964.[4,5] Congenital heart defects occur in about 50-60% of the individuals comprising of single atrium, defects of the mitral and tricuspid valves, patent ductus, ventricular septal defect (VSD), 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.[6,7]

CASE REPORT

A 25-year-old women who is lady gravida 2 abortion 1 had a spontaneous conception with 25 week 3 day gestational age, presented with breathlessness (NYHA grade 4) since 1 week for which she was referred from primary health care center to cardiac unit Shri Jayadeava Institute of Cardiovascular Sciences and Research, Mysore and after detailed cardiac evaluation found to have congenital heart disease with single atria, transitional atrioventricular (AV) canal defect, small restrictive inlet VSD left to right shunt, cleft anterior mitral leaflet (AML), moderate to severe mitral regurgitation (MR), mild tricuspid regurgitation (TR), ejection fraction (EF)-60% and sever pulmonary artery hypertension with saturation of 85% at room air, she was started on tablet torsemide 10 mg BD, tablet sildenafil 10 mg BD, tablet isolazine OD and referred to Cheluvamba Hospital attached to Mysore Medical College and Research Institute, Mysore for termination of

pregnancy as continuation of pregnancy deteriorates the mother condition. She was admitted in obstetric intensive care unit (ICU). On admission she was breathless with O_2 saturation of 85% at room air hence oxygen support was given and case was managed by multidisciplinary approach. On detailed history she had spontaneous abortion 1 year back at 1 and half month of pregnancy but no documents available told it was uneventful. There was no history of similar features in her siblings and was not a consanguineous marriage (her parents). On examination, she had hexadactyly in both upper limbs and clubbing was present, absent maxillary incisors and mandibular incisors, small chest with short ribs, cubitus valgus and genu valgum deformity present in limbs, lower limb nail dystrophy and short stature (height-133 cm) (Figures 1-5).

Per abdomen examination revealed uterus corresponds to 24-week gestational age (GA), relaxed. Relevant investigations were sent. Ultrasound had done 1 week back shows femur and humeral length less than 5th percentile. After counseling the patient attenders about the need of termination and also related mortality and morbidity, prostaglandin E1 (PGE1) induced termination of pregnancy was done. She was monitored for 48 hours and was uneventful and referred back to cardiologist for further management. At the time of referral, she was stable clinically.

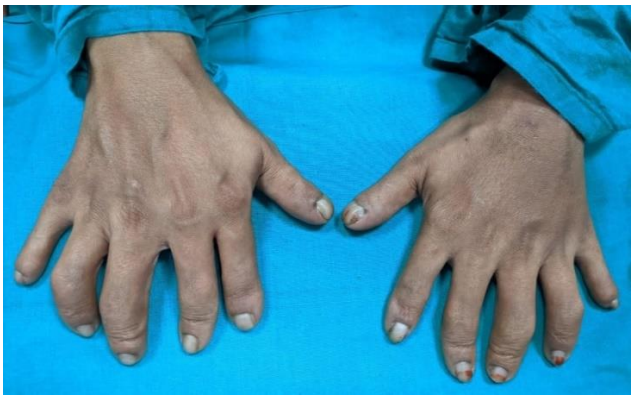

Figure 1: Hexadactyly in both upper limbs.

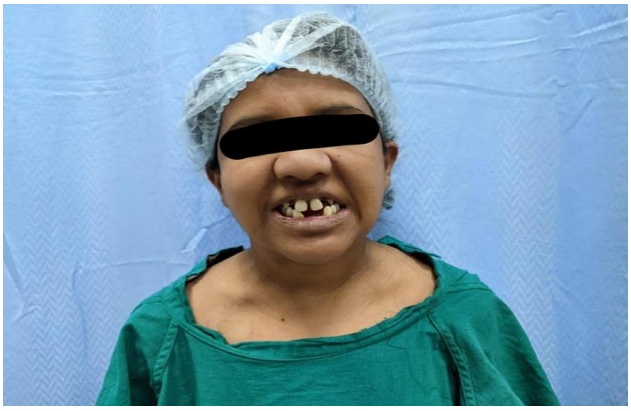

Figure 2: Congenital absence of mandibular incisors and malocclusion of teeth.

Figure 3: Cubitus valgus deformity.

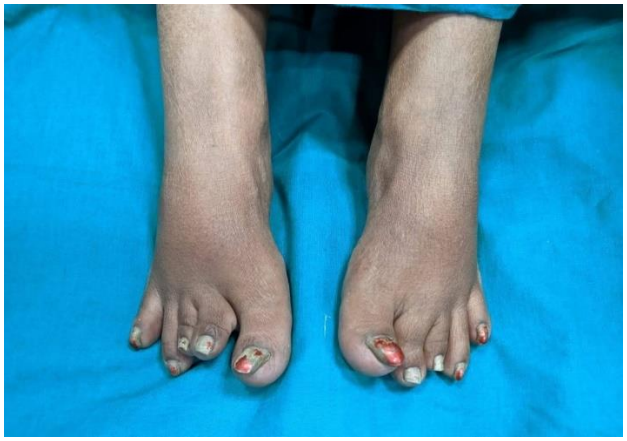

Figure 4: Dystrophic nails seen in both lower limbs.

Figure 5: Short stature.

DISCUSSION

EVC is an autosomal recessive skeletal dysplasia, characterized by short ribs, small chest, short limbs, disproportionate dwarfism, postaxial polydactyly,

congenital heart malformation and dysplastic teeth and nails. The incidence of EVC in general population is very low. The prevalence of the disease varies widely in general population of 1 in 60,000 live births in USA to 1 in 150,000 live births in European countries.[4] EVC has been included in new class of human genetic disorders called ciliopathies where underlying defect may be dysfunctional molecular mechanism in the primary cilia of cells.[8]

It is a recessive disorder found in Amish population in USA equally affecting male and female. Family history is significant in these patients with parental consanguinity is confirmed in 30% of cases.[9] Our patient was the first child of non-consanguineous marriage and normally developed parents with no significant family history. Polydactyly is constant findings in both hands, polydactyly of the feet is present in 10% of the cases. Other features include genu valgum, talipes equinovarus, pectus carinatum with long narrow chest, congenital malformation of heart is described in 50-60% of patients. The anomalies include defects of mitral and tricuspid valves, patent ductus arteriosus, VSD, atrial septal defect and hypoplastic left heart syndrome which are the leading causes of decreased life expectancy in these patients.

The disease has characteristic oral manifestation that help early diagnosis at birth or during early childhood, the most common among them include fusion of the upper lip to gingival margin resulting in the absence of mucobuccal fold, broad maxillary labial frenulum described as partial harelip, multiple small accessory frenula, ankyloglossia, and malocclusion.[10]

In the present index case, she had short stature, genu valgum, post axial polydactyly of hands, hypoplastic and deformed nails and toes, the patient had congenital heart disease in the form of single atrium. Rudnik-Schoneborn et al described about 2 patients with EVC syndrome who were 18 and 30 years old respectively.[11]

EVC needs to be distinguished from a number of closely related entities. Weyer's acrofacial dystosis is an autosomal dominant unlike EVC which is autosomal recessive, condition with many clinical features similar to EVC. However, the patients are often of normal stature and cardiac defects and thoracic dysplasia are absent.[12] Jeune syndrome, another related disorder, is an autosomal recessive condition characterized by small chest, renal anomalies and retinal degeneration.[13]

About 30% of patients die at the early age or at infancy from cardiorespiratory problems and those who survive require multidisciplinary approach for treatment, orthopaedic correction of limb deformity, surgical repair of cardiac anomaly, dental intervention if required.

CONCLUSION

It is rare congenital disorder, with high mortality in early life. One third of these patients die in infancy from cardiac and respiratory problems and those who survive multidisciplinary approach. Usually, these cases have severe cardiac anomalies which will have bad impact on her obstetric outcome. These patients have multiple anomalies which will are contraindications for pregnancy and if conceive require termination of pregnancy to prevent fatal complications. Several cases of EVC have been reported from India. To our knowledge, this is the first report of EVC patient with pregnancy.

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