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Case Report
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Ellis-Van Creveld Syndrome, a Rare Anomaly with Obvious Musculoskeletal Variations

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ABSTRACT

Introduction: we report a rare case of Ellis-Van Creveld Syndrome, which was characterized by ectodermal dysplasia, chondro dysplasia, polydactyly and congenital heart diseases. It is a rare autosomal recessive disorder, with a high mortality in early life, those who survive require multispecialty management and care. Early diagnosis and management can avoid the serious complications, morbidity and mortality.

Keywords: Ellis-Van Creveld Syndrome; Autosomal Recessive Inheritance; Mutation.

Introduction

Ellis-Van Creveld Syndrome (EVCS) is associated with ectodermal dysplasia, chondro dysplasia, polydactyly and congenital heart diseases. It is an extremely rare condition as there are only 150 cases reported globally[1,2], however the correct prevalence rate is not known. Defective development of interatrial septum is obvious in about 60% of cases[3]. But there will be no psychological and neurological deficits. This is an autosomal recessive inheritance, which is associated with mutation at the EVC1 and EVC2 gene locus at the 4p16 chromosome[4]. But these mutations are not totally accountable for the EVCS[5]. EVCS involves shorter ribs with polydactyly variety (SRP) of disorders. The management of EVCS is multidisciplinary, involves surgical correction of cardiac defects, orthopedic intervention for the bony deformities and professional dental care. The respiratory distress and cardiac anomalies in the infantile life, carry poor prognosis.

Case Report

A 31-year-old female came with swelling of both lower limbs since 3 months and abdominal distension since 2 months. On further enquiry, she gave history of exertional breathlessness and palpitations. There was no orthopnea and paroxysmal nocturnal dyspnea attacks. On examination, she had short stature measuring about 140 cms (Fig. 1A), poorly nourished. The vital signs were stable, there was no pallor and icterus. However, central cyanosis and grade 2 clubbing were present (Fig. 1B). Bilateral pitting pedal edema and abdominal wall edema were also present. However, the jugular venous pressure was not raised. The head circumference was normal. She had normal intelligence and cognition. Hypognathism (Fig. 1C) with high arched palate was noted. Ectodermal dysplasia in the form of adontia of mandibular incisors and dental enamel hypoplasia with unhealthy gums were present (Fig. 1D). She also had caries teeth. Sensorineural hearing loss on both sides were seen, which was more on the right side. Bilateral symmetrical post axial polydactyly of both hands (Fig. 2A) and feet (Fig. 2B) were noted. The bilateral cubitus valgus deformity was also present (Fig. 2C). The cardiovascular system examination revealed precordial bulge and the apex beat was localized in the left 6th intercostal space in the anterior axillary line. Right parasternal and epigastric pulsations were present. There were no thrills. On auscultation, loud S1 in the mitral and tricuspid areas and loud P2 in the pulmonary area were heard. Grade III pansystolic murmur was heard in all the areas. Respiratory system examination revealed fine crepitation in basal areas on both sides. Abdomen was distended with stretched umbilicus. Tender hepatomegaly and free fluid in abdomen were present. Central nervous system examination including higher mental functions were however normal. Her complete blood count, renal function tests, liver function tests, and serum electrolytes were within normal limits. The chest radiograph showed cardiomegaly (Fig. 3A) and the X-rays of both hands including wrists showed fusion of 5th and 6th metacarpals (Fig. 3B). Echocardiography showed single atrium (Fig. 4A) with severe mitral regurgitation (Fig. 4B), moderate tricuspid regurgitation and pulmonary hypertension.

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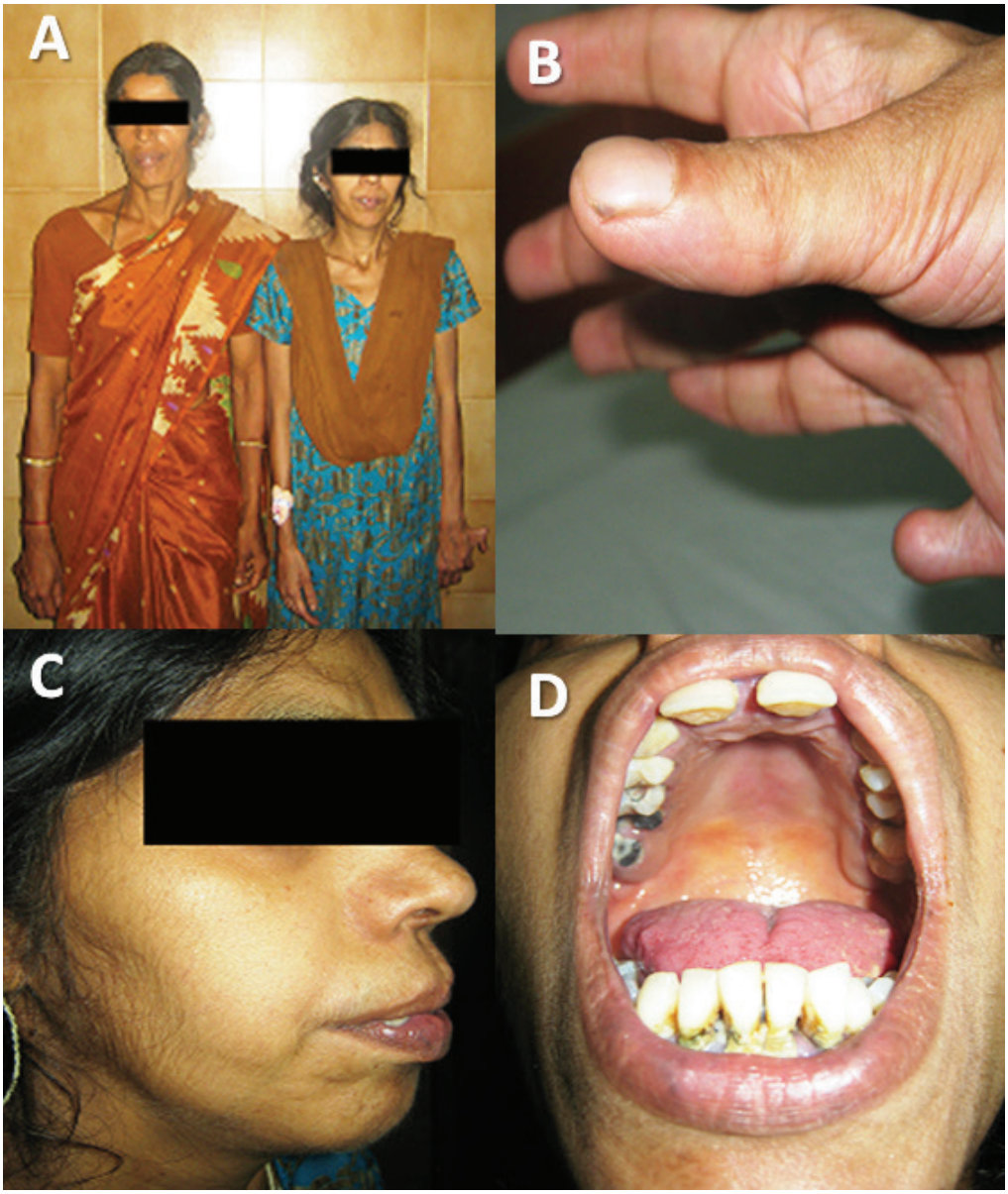

Figure 1. Morphological features of EVCS - 1A. short stature compared to sibling; 1B. grade 2 clubbing; 1C. hypognathia; 1D. dental abnormalities.

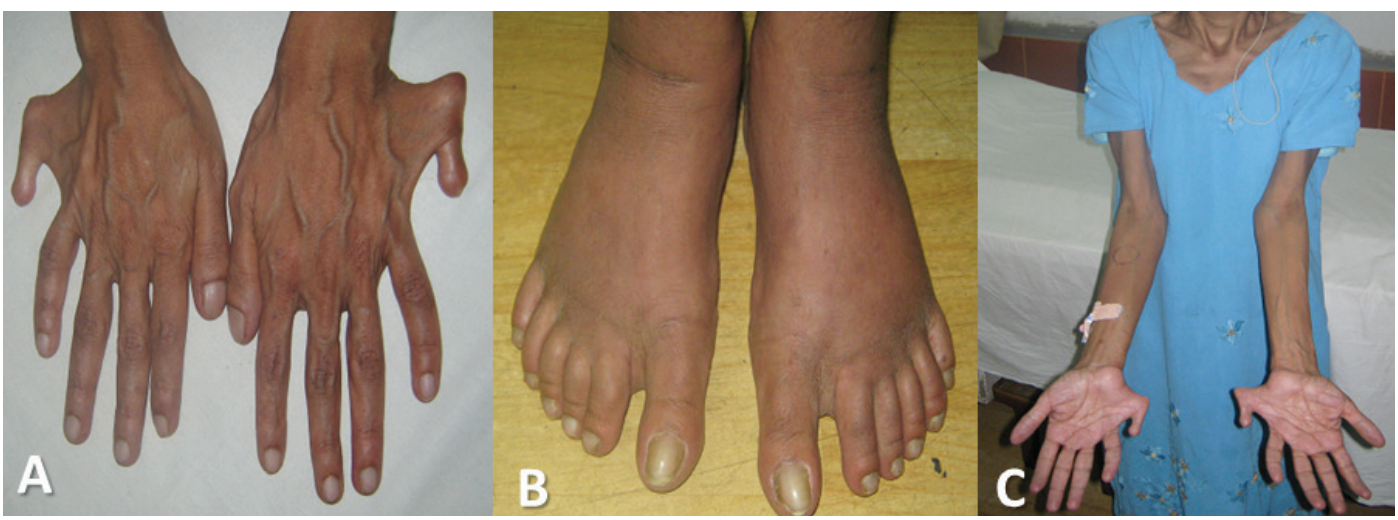

Figure 2. Skeletal deformities of EVCS – 2A. polydactyly of hands; 2B. polydactyly of feet; 2C. bilateral cubitus valgus deformity.

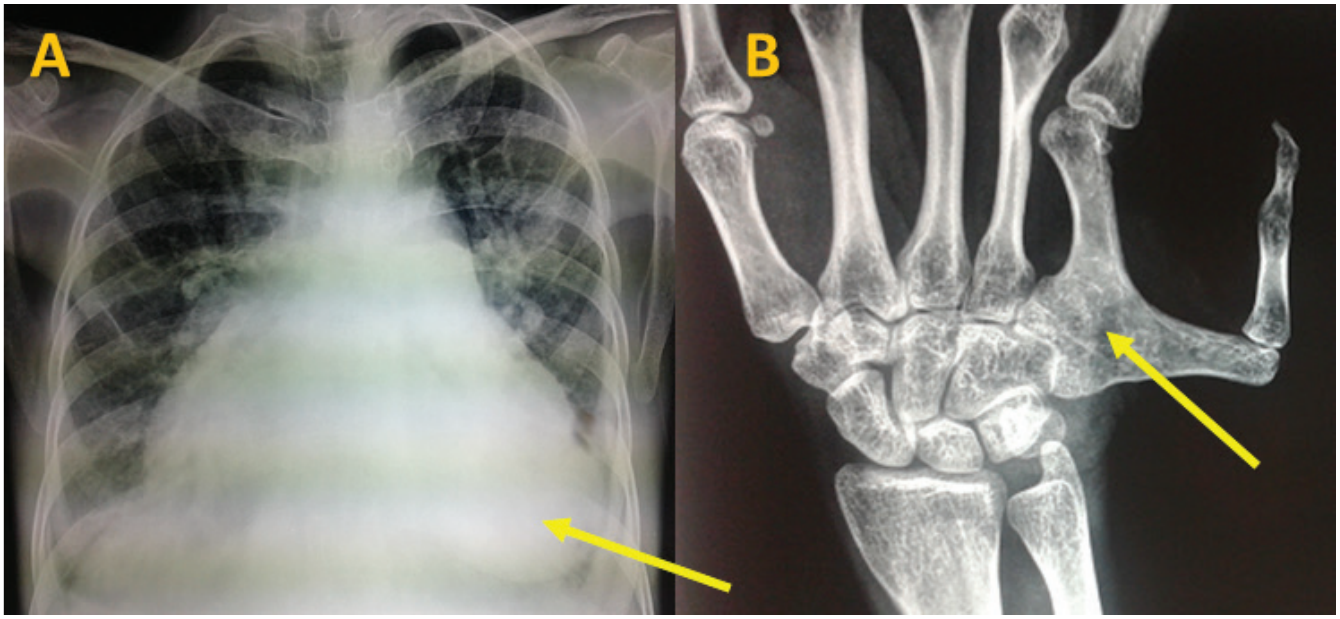

Figure 3. Radiological features of EVCS – 3A. cardiomegaly; 3B. fusion of 5th and 6th metacarpals.

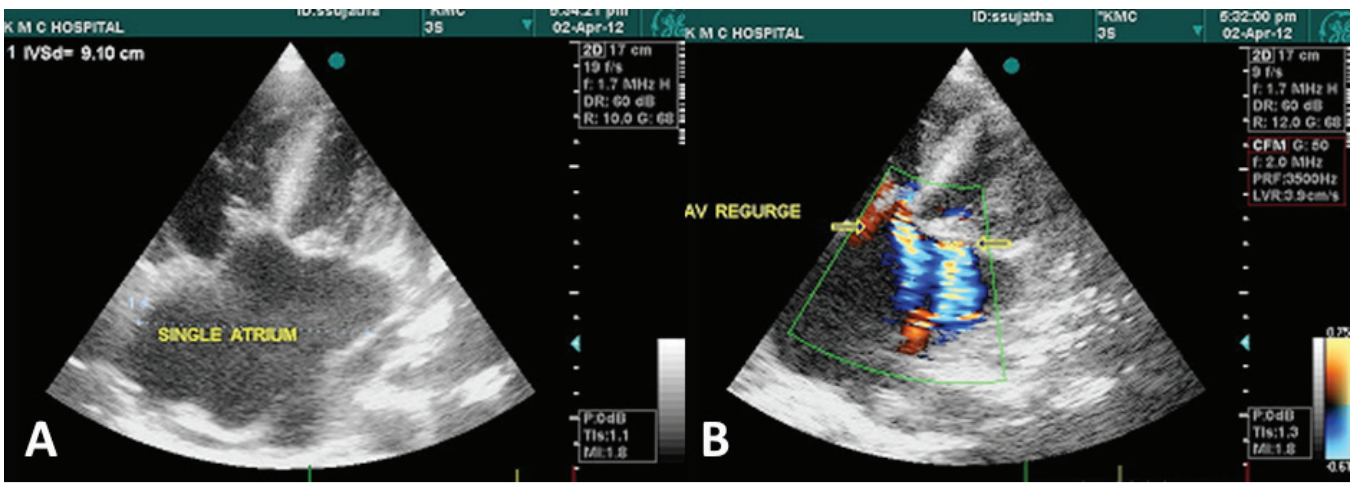

Figure 4. Echocardiography revealing – 4A. single atrium; 4B. mitral regurgitation.

Discussion

Individuals affected with EVC1 or EVC2 mutant genes have a similar spectrum of features of this syndrome and are phenotypically indistinguishable[6]. However, future research is required to figure out the other etiopathogenesis associated with this syndrome. The exact incidence is not clear, but this congenital syndrome is commonly manifested in the Amish Americans. However, the non-Amish have a prevalence of 7 in 1 million live births[7]. Polydactyly of the feet is rare and is seen only in 10% of cases[8]. Short stature is most commonly seen due to shortening in distal part of the extremity. Skeletal deformities that are usually seen are genu valgus, curvature of humerus, cubitus valgus, fusion of the medial metacarpal bones and underdeveloped ribs. The hairs are thin and sparse. Gingival hypertrophy and premature eruption of teeth, malocclusion and labiogingival adherence are the dental manifestations.

Congenital heart diseases, especially abnormalities of atrial septation (single atrium or large atrial septal defect) occurs in about 60% of cases[3]. The condition of the heart of the child depicts the life span of the child[9]. It is impossible to radiologically differentiate EVCS and other forms of chondroectodermal dysplasias such as achondroplasia, chondrodyplasia punctata and Morquio's syndrome. The differential diagnosis of EVCS should be made by considering the hypoplastic nails, cardiac defects, oligodontia and narrow chest. Prenatal diagnosis can be made as early as 18th week with IUGR, skeletal malformations (narrow chest, short limbs, hexadactyly) and cardiac defects[10]. The first-trimester nuchal translucency scan is advisable and it is associated with the higher thickness[11]. Surgical closure of cardiac variations may be required. Orthopaedic interventions for polydactyly, genu valgum and other incapacitating skeletal malformations may be performed if required. Dental intervention is a must for the tooth abnormalities and other oral manifestations.

Any major dental intervention may require antibiotic prophylaxis because of the underlying cardiac defects.

Conclusion

The clinical profile of EVCS is clearly understood until now. The presence or absence of the congenital malformations will not help in making the diagnosis of this rare autosomal recessive disorder. Early diagnosis and multispecialty care could avoid the life threatening complications.

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