Ellis-Van Creveld Syndrome with partial atrioventricular canal defect: A case report and review of literature

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INTRODUCTION

Richard W.B. Ellis of Edinburgh and Simon van Creveld of Amsterdam met in a train compartment while traveling to a pediatrics conference in England in the late 1930s and discovered that each had a patient with the specific similar pathology because of chondroectodermal dysplasia and named it Ellis van Creveld Syndrome. It is an autosomal recessive syndrome with disproportionate dwarfism, postaxial polydactyly, ectodermal dysplasia, a small chest, and a high frequency of congenital heart defects. Other features include partial harelip and multiple frenulae in the lips; short ribs and narrow chest; genital abnormalities like epispadias, hypospadias and cryptorchidism; low iliac wings with spur-like

Amish population of Pennsylvania in USA but also occur in non Amish population with the prevalence around 7/1,000,000 live births. Here we report a case of 15 year old Indian female born to a consanguineous marriage with the classical features of Ellis Van Creveld Syndrome diagnosed with a partial AV canal defect having a relatively asymptomatic childhood. A Partial AV canal defect is an uncommon cardiac malformation, and yet it is commonly found in patients with the EVC. We operated on her with septal patching and anterior mitral leaflet cleft repair with smooth recovery.

Key Words: EVC- Ellis Van Creveld syndrome; PA VCD-Partial AV canal defect; AML-Anterior Mitral Leaflet

CASE REPORT

A 15 years young female presented to us with progressive exertional dyspnea and easy fatigability of six months duration with no history of orthopnea or PND. She had no significant illness in the past except that she was born with short limbs, six fingers in each hand and six digits in each foot [Figure 1A]. She also has a history of frequent respiratory tract infections. She had short stature which was apparent from two years of age. Her school performance was below average. She attained menarche at the age of ten years and having normal menstrual cycles once in a month since then with no h/o any other bleeding tendencies. On examination, she was found to have a short stature with a height of 115 cm. Her younger sister also, was having short stature. The finger and toe nails were small and brittle. Oral cavity examination revealed absent incisors and the rest were natal teeth. The patient had knock knees [Figure 2A] with pectus excavatum. Examination of cardio-respiratory system revealed narrow chest and had a prominent diastolic thrill at the apex, a loud S1 with a low-pitched grade 4/6 mid-diastolic rumbling murmur and a pansystolic murmur in the mitral area. The S2 was widely split, fixed with a loud P2. There was a grade 3/6 ejection systolic murmur over the left second intercostal space. Other systems were normal. All blood investigations are normal. Skeletal radiology of upper limb revealed short metacarpals, ulnar thick 5th metacarpal, ulnar polydactyly [Figure 1B] and X-ray foot showed short metatarsals with six toes [Figure 2B]. Radiological examination of the lower limb revealed depressed and flattened lateral compartment of upper end ofibia on both sides giving rise to genu valgum. X-ray skull showed craniofacial disproportion [Figure 3A]. Chest X-ray showed cardiomegaly. Ultrasound abdomen was normal. Electrocardiogram showed normal sinus rhythm, first degree heart block with biatrial enlargement. Echocardiography showed partial AVC-D-large ostium primum atrial septal defect amounting to single atrium, two separate atrioventricular valves with moderate mitral regurgitation, cleft in AML-right atrium and right ventricle dilated, mild pulmonary artery hypertension. Catheterization study showed classical Goose Neck deformity due to elongated left ventricular outflow tract in the left ventricular angiogram. The saturation run showed significant step up in the

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Abbreviations- EVC- Ellis van creveld syndrome

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right atrium with QP/QS-5.65. Mean pulmonary artery pressure was recorded as 30 mm Hg. We operated on her with pericardial patch closure of ostium primum atrial septal defect and repair of cleft in AML with 5-0 prolene through right atriotomy on cardiopulmonary bypass after arresting the heart under GA [Figure 3B]. Postopt recovery was uneventful. Echo follow up showed that the patient’s symptoms have reduced with increased tolerance for work.

Six-fingered dwarfism ('digital integer deficiency') was an alternative designation used for this condition when it was being studied in the Amish however, has been found offensive by some, apparently not because of the term, as a 'freakish' labelling [3]. For this reason, 6-fingered dwarfism has been removed as an alternative name for this entry. This leaves Ellis–van Creveld syndrome. Other genitourinary anomalies like renal agenesis, renal tubular dilation, nephrocalcinosis, megaureters, vulvar atresia. 50% of patients with Ellis Van Creveld syndrome usually die in infancy due to recurrent respiratory infections. Those without any cardiac abnormality may have a normal life expectancy. The patient having a complete atrioventricular canal defect with EVC has survived till 19 years without any significant pulmonary artery hypertension [10]. There are only few reports of cardiac surgical repairs done for this condition. Our patient was born to a consanguineous marriage with similar EVC syndromic features in her younger sister. After diagnosis of PAVCD we investigated the patient in a multidisciplinary way and then operated as a routine cardiac surgery with no intraopt or postopt complications.

**CONCLUSION**

Ellis van Creveld syndrome, though rare in Indian population, but with a multidisciplinary approach with involvement of dentist, psychologist, paediatrician, urologist, orthopedician, pulmonologist, cardiologist, cardiac surgeon and occupational therapist management seems to be easy and simple. Early diagnosis and treatment of cardiac abnormality in EVC syndrome patients will definitely decrease the morbidity and mortality with improved quality of life.

Ethical approval: All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

**REFERENCES**