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Case Report

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A RARE CASE REPORT ON ELLIS VAN CREVELD SYNDROME

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ABSTRACT

Ellis Van Creveld syndrome (EVC) is a rare genetic disorder having autosomal recessive inheritance affecting, which may clinically present as small stature, short limbs, fine sparse hairs, hypoplastic fingernails, and enamels. This syndrome consists of characteristic features such as bilateral postaxial polydactyly, chondroectodermal dysplasia congenital heart defects and hypoplastic nails and teeth. We report a case of a 17year old male presenting the typical features of this syndrome.

KEYWORDS: Ellis Van Creveld Syndrome, polydactyly, chondroectodermal dysplasia.

INTRODUCTION

Ellis Van Creveld Syndrome was described in 1940 by Richard W.B. Ellis and Simon Van Creveld as a rare autosomal recessive disorder. It is a genetic defect located in chromosome 4p16.[1] It is caused by mutation in a known homologous gene EV2, located in a head-to-head configuration with parental consanguinity in about 30% of the cases.[2] The severity of the disease varies from person to person. The highest rate of the condition is seen among the old order Amish Population Lancaster Country, Pennsylvania. It is fairly rare in general population.[3] The disease can be diagnosed at any age, even during pregnancy.[4] The presence of a variety of oral manifestations, such as fusion of upper lip to gingival margin, presence of multiple frenula, abnormally shaped microdontic teeth and congenitally missing teeth requires multidisciplinary dental treatment with consideration for the high incidence of cardiac defects in these patients.[5]

CASE REPORT

A 17 year old male patient was admitted in NIIMS hospital Neyyatinkara for the complaints of dental problem, multiple frenula and polydactyly. He had a h/o short status, polydactyly, small nails, bat ears, multiple frenula and dental problems and admitted in Sree Chithra Medical College Trivandrum at the age of 3. No h/o cyanosis, recurrent RTI, family h/o – elder sister had the similar disease and died at the age of 3 year, mild neurodevelopmental delay. His lab investigation includes HBsAg negative. Echo-normal, Blood group of AB+. His surgical findings include Inter Atrial Septum absent except for the superior rim. Both AV valves seem through the defect. All PV draining to LA, coronary sinus draining to LA. Tiny cleft in AMI, not causing any significant MR.

His treatment includes an antibiotic Cefuroxime 100mg BD, Tab.lasix 5mg PO BD, Tab.Aldactone 6.25mg PO BD, Cap.Idicin 12.5mg PO TDS and Cap.Fesovit OD.

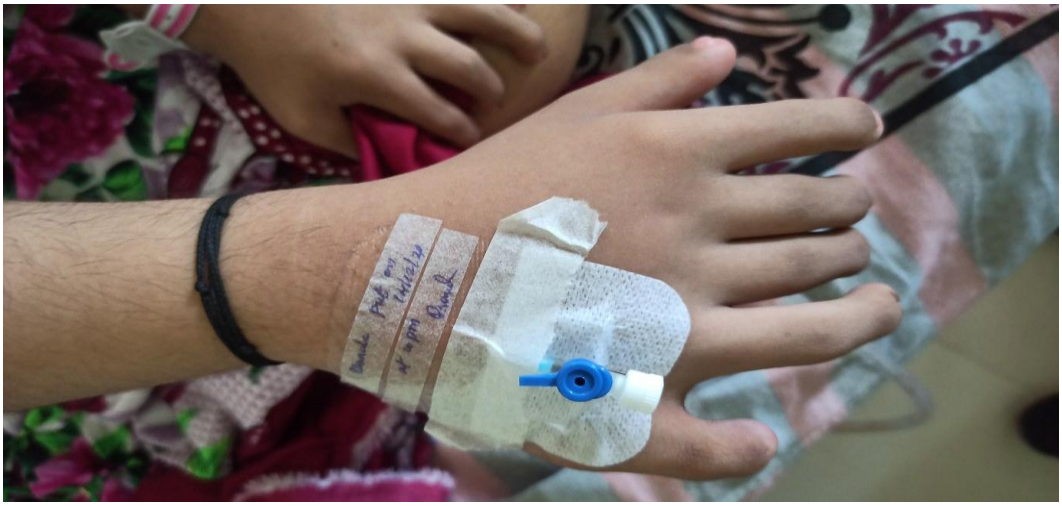

Fig. 1: Showing polydactyly of the hand.

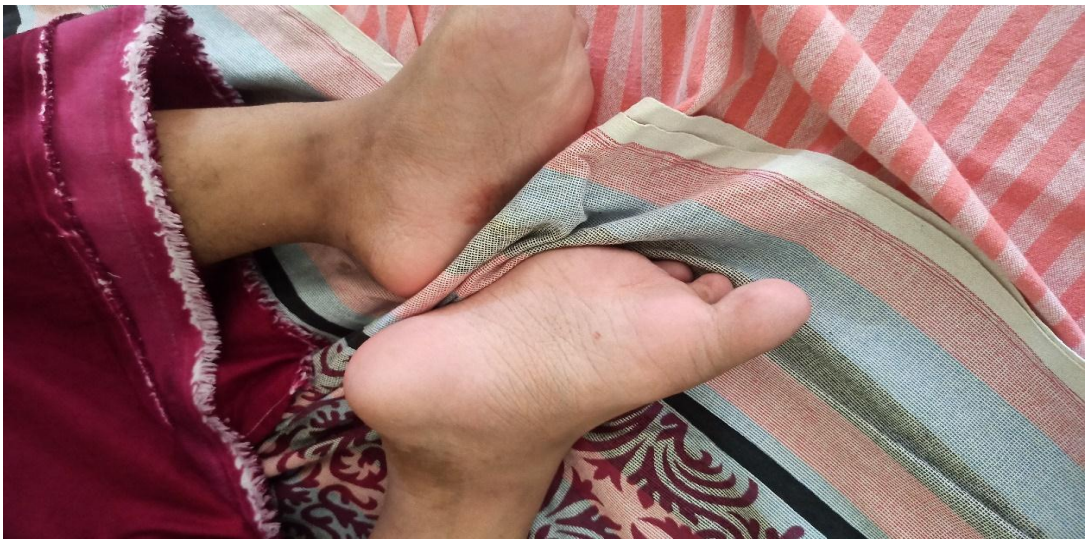

Fig. 2: Showing Polydactyly of the foot and small status.

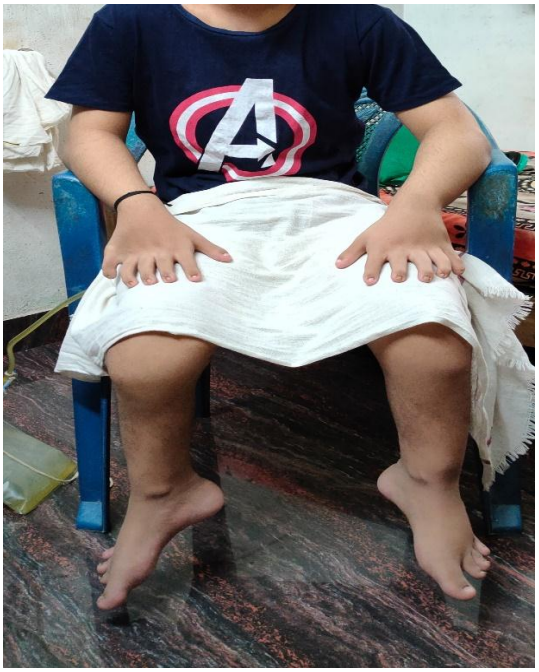

Fig. 3: Showing small stature, short limbs and hypoplastic fingernails.

DISCUSSION

Ellis Van Creveld Syndrome also known as chondroectodermal dysplasia and is genetic disorder with autosomal recessive inheritance and parental consanguinity.[2] seen commonly in Amish population of Pennsylvania USA. Our patient is the brother of genetically inherited sister.

The most consistent clinical feature is chondrodystrophy due to defect in ossification affecting tubular bones resulting in shortened long bones of the limbs resulting in shortened long bones of the limbs especially in the distal and middle segments resulting in acromesomelic dwarfism.[3] The other features include polydactyly having wide hands and feet, sausage shaped fingers and dysplastic figure nails. Our patient had polydactyly in upper and lower lip. Other features include difficulty in walking. The disease has characteristic oral manifestations that help early diagnosis at birth or during early childhood.[5] The most common among them include fusion of the upper lip to the gingival margin resulting in the absence of mucobuccal fold, broad maxillary libal frenum described as partial harelip, multiple small accessory frenula, conical microdontia teeth and enamel hypoplasia.[5] Our patient had all these findings.

At the beginning of 4.5 months of gestation (18^{th} week), USG (ultrasonography) and clinical examinations can help diagnose this syndrome.[3] One important and most common clinical finding observed in this syndrome is chondrodystrophy. In this case, most commonly, long bones of extremities like the femur (sometimes patella) are involved giving rise to a defect in ossification.[1] We usually observe that the thorax is sunken with funnel chest (pectus excavatum), inward curving of the lower back (lordosis), and inward turning of the lower extremities (genu valgum). Hairs are usually less and fine. The appearance of multiple digits (polydactyly), broad hands and feet, dysplasia of nails of fingers, and sausage-like fingers typically point towards the diagnosis of EVCS being very particular features for this syndrome.[1]

The beneficial and functional management of this syndrome should consist of a team comprising of a geneticist, oral and maxillofacial surgeon, pedodontist, prosthodontist, paediatrician, pediatric cardiologist, urologist, orthopaedician, psychologist, pulmonologist and pediatric neurologist.

CONCLUSION

EVCS is a very rare and still an incurable condition but there could be several kinds of management for this syndrome. The presence of several rare cardiac anomalies like patent ductus arteriosus (PDA), ASD, and three pulmonary veins in our case is rare in association with this syndrome. This condition can be surgically tackled in one sitting using an autologous pericardial patch along with ligation of PDA and ASD patch closure which will improve the condition and lifestyle of the patient. The extrapulmonary vein can be ligated. This case report will help readers gain knowledge about the rare findings of this particular syndrome and help medical professionals prognose, diagnose and treat patients in an effective and faster manner.

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Nil

CONFLICTS OF INTEREST

The authors have the required patient consent forms, on which the patients have agreed to participate in the study and be represented in the corresponding publication. Although the patients are aware that the writers would take precautions to keep their names secret, anonymity cannot be guaranteed.

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