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ELLIS VAN CREVELD SYNDROME – A RARE CASE REPORT IN A NEONATE

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ABSTRACT

Background : Ellis-van Creveld (EVC) syndrome is a genetic disorder with autosomal recessive transmission with mutation in EVC and EVC2 gene .It clinically presents with polydactyly, short stature, fine sparse hair, hypoplastic fingernails, natal teeth, enamel hypoplasia , hypodontia, and malocclusion. Heart defects, especially abnormalities of atrial septation, have been found in about 60% of cases. **Case Presentation:** In this article, we present a rare case of Ellis Van Creveld syndrome in a 4 day old male neonate with suckling difficulty, polydactyly and uvular shift . **Conclusion:** It is important to diagnose such syndromes early in life so that they can be provided with symptomatic care and support as well as appropriate surgeries when required. **Summary** Here, we present a case report of a 4 day old male neonate who was brought by his relatives with difficulty in sucking milk . On examination, he had polydactyly in both hands and left leg . On oral examination, he had labiogingival frenum hypertrophy and uvular shift to right side . USG brain showed grossly dilated lateral ventricle. He was advised to be on regular follow up for monitoring the development of any skeletal abnormalities and for oral surgeries.

INTRODUCTION

Ellis-van Creveld (EVC) syndrome is an autosomal recessive chondroectodermal dysplasia, first reported by McIntosh in 1933 and described and defined as EVC syndrome by Richard W.B. Ellis of Edinburgh and Simon van Creveld of Amsterdam in 1940.[1] This syndrome belongs to a group of diseases called ciliopathies, where a mutation in two adjacent genes on chromosome 4 leads to abnormalities in the primary cilia .The genes are EVC and EVC2, which play a role in ciliary development .[2]

It is also known as mesoectodermal dysplasia and is more predominantly found in the Amish community in Pennsylvania, United States. It has now been described to affect all races though its exact prevalence is unknown.[3]

Case Report

A 4 day old male neonate of Indian origin reported to our institution after caesarian delivery with the chief complaint of difficulty in sucking milk.

He is post term with a birth weight around 2.75 kg and is the second child of a non-consanguineous marriage with the elder sibling having no congenital abnormality. On general physical examination, the patient was responsive with normal cry. Examination of the hands and feet revealed bilateral postaxial polydactyly in both hands (Fig1& 2) and left foot (Fig3). There was an outward bending of the left knee (genu varum) .Hair and nails were normal.

Intra-oral examination revealed presence of conical lower central incisors as natal teeth with multiple frenulum and labiogingival frenum hypertrophy (Fig 4) with uvular shift towards the right side which lead to sucking and swallowing difficulties. (Fig 5)

His routine investigations were normal. He was advised Cardiology, Plastic surgery, Pediatric, Ophthalmology and Dental consultations for his symptoms. Echocardiography showed a patent foramen ovale with a left to right shunt. USG brain was done which showed grossly dilated lateral ventricle for which MRI was advised later in life. USG abdomen was normal. His ophthalmological and skeletal evaluation was normal at the time of presentation.

Since our hospital has a government set up, a genetic mutation analysis could not be done due to the lack of availability. He was advised regular follow ups to look for the development of skeletal malformations and surgeries for his oral deformities but was lost to follow up.

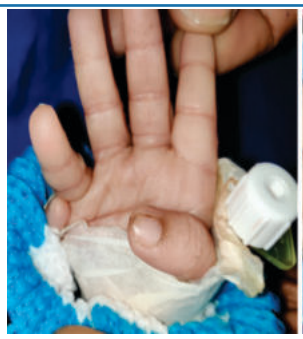

Fig 1: shows polydactyly – right hand

Fig 2: shows polydactyly – left hand

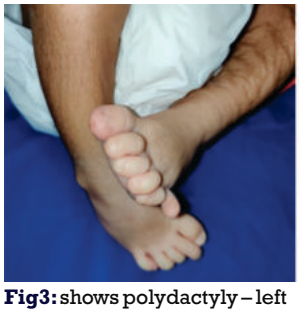

Fig3: shows polydactyly – left leg

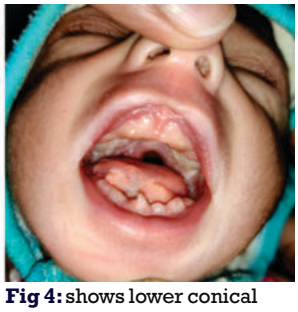

Fig 4: shows lower conical tooth with labiogingival frenum hypertrophy

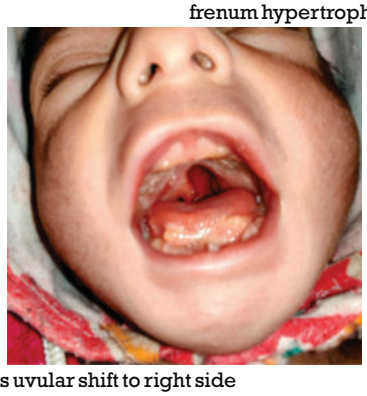

Fig5 : shows uvular shift to right side

DISCUSSION

EVC syndrome is an ectodermal dysplasia and the genes responsible are EVC 1 which codes for EVC protein and EVC 2 which codes for limbin both of which leads to cilia

dysfunction. The most important clinical features include polydactyly which can be unilateral or bilateral usually affecting the hands and sometimes feet, disproportionate small stature, hidrotic ectodermal dysplasia affecting the nails, hair, and teeth. Bilateral polydactyly is seen in only 10 % cases

The characteristic oral manifestations which help in early diagnosis during childhood include conical teeth, labiogingival fusion, natal teeth, premature eruption, and absence of mucobuccal fold, enamel hypoplasia, multiple labial gingival frenulae and short upper lip.[4]

Congenital cardiac malformations like defects of mitral and tricuspid valves, patent ductus, single atrium, ventricular septal defect and atrial septal defect have been reported in about 60 % of the cases.[5]

Skeletal malformations include genu valgum, lumbar lordosis, talipes equinovarus, defects of the femur and humerus and pectus carinatum can be associated. Other uncommon anomalies which can be seen are fissured tongue, congenital stridor, strabismus, congenital cataracts, dysplasia of the kidney, urinary stones, cryptorchidism and epi- and hypospadias.[6]

It can be diagnosed prenatally by ultrasonography after the 18th gestation week.[7] Diagnosis at birth can be made clinically by the typical symptoms supported by radiography, ECG, and echocardiography.[8] The definitive diagnosis is by molecular diagnostic techniques, which is based on homozygosity for a mutation in the EVC and EVC2 genes by direct sequencing.[9]

The differential diagnosis of the syndrome includes Saldino-Noonan syndrome, Verma-Naumoff syndrome, Beemer-Langer syndrome, Majewski syndrome, Weyers syndrome, Jeune Dystrophy and McKusick-Kaufman syndrome

Treatment is usually symptomatic. A multidisciplinary approach involving Plastic Surgeon, Dentist, Cardiologist, Orthopedist, Physiotherapist, etc. are required. If a cardiac abnormality is detected, a surgical or non-surgical management of the defect should be done. Dental treatment is usually done early in life like treating cleft lip, and teeth alignment. No definitive cure have been found [10]

Even though DNA mapping studies could not be done, the presence of typical oral manifestations like natal teeth, multiple labial gingival frenulae and labiogingival adherence helped us to distinguish it from several other similar syndromes. Our patient also had difficulty in sucking milk due to his oral malformations which can itself lead to failure to thrive and therefore surgical correction should be done as early as possible. All the previous case studies published on EVC syndrome have been diagnosed and reported at a later age. We have been able to diagnose the syndrome at the early stages of neonatal period and further follow-ups are required to look for the development of skeletal abnormalities and disproportionate growth.

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