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Ellis Van Creveld Syndrome: An Unusual Presentation at Birth

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1. Abstract

The Ellis-van Creveld Syndrome, also known as Chondroectodermal dysplasia, is a skeletal and ectodermal dysplasia which is characterized by the narrow chest, short ribs with congenital heart defects like ASD and polydactyly. It is an extremely rare condition. An unusual case of Ellis -van Creveld Syndrome is described, which was diagnosed by typical clinical features like narrow funnel shaped chest, polydactyly and congenital heart disease like single atrium. Unusual features being, presence of natal tooth, single atrium instead of ASD (atrial septal defect) and talipes equinovarus deformity. Child presented soon after birth (2 hours) with respiratory distress and requirement of supplemental oxygen. Genetic Confirmation could not be done because of short stay no family history of this illness found in family.

2. Introduction

The Ellis-Van Creveld syndrome, also known as Chondroectodermal dysplasia is a skeletal and ectodermal dysplasia which is characterized by the narrow chest, shot ribs with congenital heart defects like ASD with polydactyl [1]. The syndrome was first described by Richard W. B. Ellis (1902-1966) and Simon van Creveld (1895–1971). It is an autosomal recessive trait caused due to mutation of EVC1 or EVC2. We describe below an atypical case of Ellis Van Creveld syndrome with single atrium, talipes equinovarus, natal teeth and polydactyly [2].

3. Case Report

A term baby was bought at 2 hours of life, a product of normal vaginal delivery at home, to a second gravida un-booked mother born out of nonconsanguineous marriage. Child presented with respiratory distress and blueness of the body since birth. The pregnancy was uneventful with no adverse peri-natal factor. There was no history birth anoxia.

On examination at admission baby appeared sick with heart rate of 154/min, respiratory rate 74/min with severe subcostal retractions. SpO2 at room air was 82% with obvious cyanosis, with 5 litre of O2 delivery by Hood the SpO2 became 97%. The weight recorded was 2800 grams, length 43 cm and head circumference 34 cm. The patient had dysmorphic features in form of short limbs, bell shaped chest (Figure I) with polydactyly (Figure III), right side plantar eversion (Figure V) and natal teeth (Figure IV).

Figure I: Neonate with narrow chest and equinovarus talipes

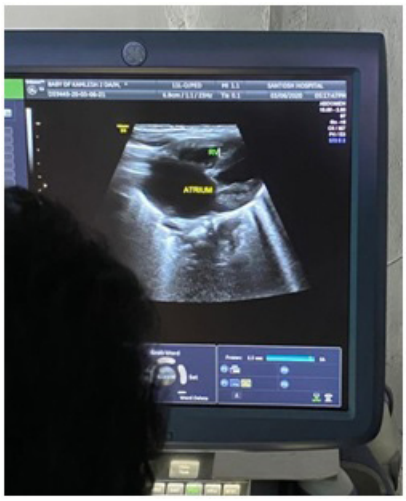

Figure II: Absence of interarterial septum

Figure III: Bilateral Polydactyly in EVC patient

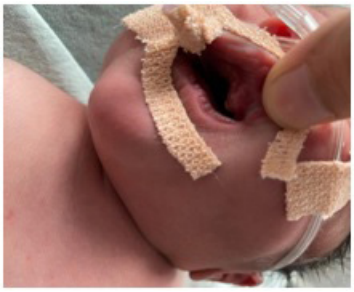

Figure IV: Anterior view of mouth showing Natal Teeth

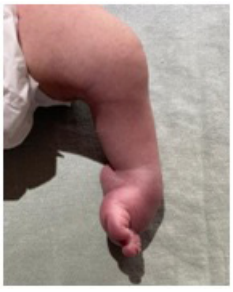

Figure V: Right limb showing Equinovarus Talipes

The investigation were carried out following admission, complete blood count being normal. The C- reactive protein was negative. The serum calcium levels were 7.9 mg/dl with potassium levels being 5.9 mEq/L. Ultrasonography of cranium didn't show any evidence of hydrocephalus or intracranial bleed. Echocardiography of heart subcostal view was suggestive of Single Atrial chamber.

4. Discussion

Ellis-van Creveld syndrome also known as chondroectodermal dysplasia [3], is characterized by chondrodysplasia, polydactyly, ectodermal involvement and congenital heart defects. The skeletal dysplasia presents with short limbs specially in middle and distal segment. It may be accompanied with polydactyly of hand and at times of the feet too. Close differential diagnosis is Jeune's Asphyxiating Thoracis Dystrophy which does not have heart defects [4].

4.1. The Above Mentioned Case Showed All Features of the Syndrome Except for Nail and Hair Dysplasia and Presented with Single Atrium

Ellis Van Creveld is a rare autosomal syndrome which can be diagnosed prenatally by intrauterine growth restrictions, skeletal malformation and cardiac defects on ultrasonography or by using chorionic villi or amniotic fluid study in a previously affected sibling. Clinical diagnosis is done based on the symptoms and manifestations. The definitive diagnosis is by the molecular study on homozygosity for mutation in the EVC and EVC2 genes [5]. Due to short stay in the hospital the genetic analysis could not be done for the neonate hence, based on clinical spectrum we came to diagnosis of the syndrome.

5. Prognosis

Till date there are no series of systematic follow up for EVC, but the prognosis is related to respiratory condition and heart defect. Ellis Van Creveld syndrome has high mortality in early life due to cardiac and respiratory problems. A multidisciplinary team approach is always advised which includes a cardiologist, a pediatrician, an orthopedic, a prosthodontist, an oral and maxillofacial surgeon, an orthodontist for management and rehabilitation of such patients.

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