

Ellis-Van Creveld Syndrome With Hypospadias And Undescended Testis: A Rare Case Report in An Indian Paediatric Patient

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I. Introduction

Ellis-van Creveld syndrome (EVCS) is also known as chondroectodermal dysplasia or mesoectodermal dysplasia, is a rare autosomal recessive disorder, caused by mutations in the EVC(EVC1) and EVC2 gene (4p16), mapped to the short arm of chromosome 4.¹ The disorder was described by Richard WB Ellis (1902–1966) of Edinburgh and Simon van Creveld (1895–1971) of Amsterdam.² EVCS presents with the characteristic tetrad of: (1) Disproportionate dwarfism with short limbs and exceptionally long trunk, (2) bilateral postaxial polydactyly of the hands, (3) dystrophic nails, hypodontia and malformed teeth, (4) congenital cardiac malformations occur in 50 to 60% of cases, most common being the interseptal defect.² In most parts of the world, Ellis-van Creveld syndrome occurs in 1 in 60000 to 200000 newborns. It is difficult to estimate the exact prevalence because the disorder is very rare in the general population. This report describes a case of Ellis-van Creveld syndrome in a 11 months old male child with the tetrad of principal features.

II. Case Report

A 11 months old male child reported to the Department of Paediatrics and Neonatology, RIMS, Ranchi with the chief complaint of fever, cough and difficulty in breathing. Patient was the second child of healthy parents of non-consanguineous marriage.

Detailed history revealed repeated lower respiratory tract infections in the past requiring hospitalisation. Antenatal, natal and neonatal histories were non contributory. No significant family history. On general examination the weight was 7.4kg, length 68cm, head circumference 43.2cm, narrow chest, protuberant abdomen, generalised hypotonia, short distal extremities[Fig.1], 2 small peg shaped teeth in the lower jaw present since birth[Fig.2], polydactyly in all 4 limbs[Fig.3.4] with small dysplastic nails. He also had hypospadias and undescended testis. Based on the clinical and radiographic findings, the patient was diagnosed to have EVC syndrome.



Fig . 1 Whole body photograph showing Disproportionate dwarfism with short limbs and exceptionally long trunk.



Fig. 2 Facial photograph showing natal teeth.



Fig.3 Polydactyly of upper limb.



Fig. 4 Polydactyly in the lower limbs.

III. Discussion

Ellis Van Creveld Syndrome also known as chondroectodermal dysplasia is genetic disorder with autosomal recessive inheritance and parental consanguinity.^{1,3,4} seen commonly in Amish population of Pennsylvania USA which does not show any gender predilection.³ The diagnosis can be made as early as 18th week of gestation by ultrasonography when the increased nuchal translucency is evident.^{5,7,8} and later by clinical examination after birth.^{2,4,5}

The most consistent clinical feature is chondrodystrophy due to defect in ossification affecting tubular bones resulting in shortened long bones of the limbs especially in distal and middle segments resulting in acromesomelic dwarfism.^{4,5,8} The other features include polydactyly usually bilateral postaxial hexadactyly most often seen in upper limbs on ulnar side and involves lower limb in 10% of cases. They also have wide hands and feet, sausage shaped fingers and dysplastic fingernails.^{3,8} Our patient had hexadactyly in both the upper and lower limbs with no syndactyly. Other features include genu valga, curvature of the humerus, talipes equinovarus, talipes calcaneovalgus⁵ and pectus carinatum with a long narrow chest.^{3-5,8} Congenital heart malformations are described in a 50–60% of patients. The anomalies include defects of the mitral and tricuspid valves, patent ductus arteriosus, ventricular septal defect, atrial septal defect and hypoplastic left heart syndrome which are the principal causes of decreased life-expectancy in these patients.^{4,8,9}

Genitourinary abnormalities are seen in about 22% of the cases and include vulvar atresia, megareters, nephrocalcinosis and renal agenesis.^{4,8} Several inconstant additional clinical findings are described, including strabismus, epi- and hypospadias, cryptorchidism and thoracic wall and pulmonary malformations.^{4,7} Exceptionally, hematological anomalies such as dyserythropoiesis and perinatal myeloblastic leukemia have been reported.^{4,7} The cognitive and motor development are normal with occasional CNS anomalies and hydrocephaly.^{8,9}

The definitive diagnosis is molecular based on the homozygosity for a mutation in the EVC 1 and/or EVC 2 genes by direct sequencing. However the genetic mutations are seldom required for the clinical diagnosis as gene mutations is positive in only 2/3rd of patients. Due to the lack of availability of genetic studies the diagnosis was achieved clinically based on the observation of the symptoms and manifestations as described and with the aid of additional tests such as radiology, laboratory and cardiac function.^{2,4,8}

Differential diagnosis includes other short rib polydactyly syndromes like Weyers acrodistal dysostosis (Curry-Hall syndrome), asphyxiating thoracic dystrophy (Jeune syndrome), achondroplasia, chondroplasia punctata, orofaciocaudal syndromes and Morquio's syndrome.^{4,5,7,8}

To conclude a multidisciplinary team approach is always advised which includes paediatrician, paediatric neurologist, clinical geneticist, cardiologist, pulmonologist, orthopaedician, urologist, psychologist, pedodontist, oral and maxillofacial surgeon for suitable diagnosis, management and rehabilitation of such patients.

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