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Case Report
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Ellis-Van Creveld Syndrome in Indian Child

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Abstract

Ellis-van Creveld syndrome is a rare genetic disorder. It was first described by Richard Ellis and Simon van Creveld in 1940. The orofacial manifestations include multiple gingivolabial musculofibrous frenula, dental anomalies, hypodontia and malocclusion. Chondrodysplasia, polydactyly, ectodermal dysplasia and congenital heart defects also has been reported. Here we report such a case in an Indian child.

Keywords: Ellis-van Creveld syndrome; Dysplasia chondroectodermal; EVC1 and EVC2 genes

Introduction

A 59 days old male child was reported to our department with the complaint of a tooth in the lower jaw since birth, continuous crying, and refusal to suck milk (Figure 1).

Oral examination revealed one crown of the teeth in the mandibular anterior region (Figure 2), whitish. No mobility was noted. The crown size and the gingiva were of normal appearance. The mother reported that it was present at the time of birth itself.

Our patient was the second son of non-consanguineous marriage and normally developed parents (Pedigree analysis Figure 11). There was no history of any familial diseases. The patient's mother was taking steroid medication for bronchial asthma in the earlier months of pregnancy. She reported that in the third antenatal scan itself, shortness of long bones were suspected soon after delivery, the baby developed cough and dyspnea and was hospitalized. The patient's birth weight was 3.2 Kg. Head circumference- 34.4 cm. At the time of reporting his weight was 4.04 kg, Height: 36.4 cm, Length: 55 cm.

On examination, the patient was found to be having dysmorphic facies, epicanthic fold, polydactyly of right and left hands, narrow chest, natal tooth in lower posterior region, short upper and lower limbs, rhizomelia, etc. (Figures 4-10). Our patient had hexadactyly in both the upper and lower limbs with no syndactyly (Figure 4, 5).

Based on the clinical and radiographic findings of the dental and medical examination, the patient was diagnosed to have EVC syndrome. The patient was referred to the Department of Pediatrics for further evaluation. The patient underwent color Doppler echocardiography and found to be having atrial septal defect, small ventricular septal defect, and mitral regurgitation, and right pulmonary artery stenosis. Genetic consultation was done at the pediatric genetics department and the final diagnosis was Ellis-van Creveld syndrome.

Discussion

Syndrome Ellis-Van Creveld (EVC) was described in 1940 by Richard W.B. Ellis and Simon Van Creveld as a rare autosomal recessive disorder due to a genetic defect located in chromosome 4p16.1e3 they termed it as dysplasia chondroectodermal [1]. Syndrome consists of bilateral postaxial polydactyly of the hands, chondrodysplasia of long bones resulting in Acromesomelic dwarfism, ectodermal dysplasia affecting nail and teeth, and less often congenital malformations [1]. The syndrome has been included in a new class of human genetic disorders called 'Ciliopathies' recently, where the underlying defect may be a dysfunctional molecular mechanism in the primary cilia of cells [2]. Syndrome has an autosomal recessive inheritance. In 30% of cases, parental consanguinity will be there [3,4].

Ellis-Van Creveld syndrome is associated with abnormalities (mutations) in two genes on the number 4 chromosome called EVC1 and EVC2. These gene mutations result in the production of abnormally small EVC1 and EVC2 proteins. Some affected individuals do not have mutations

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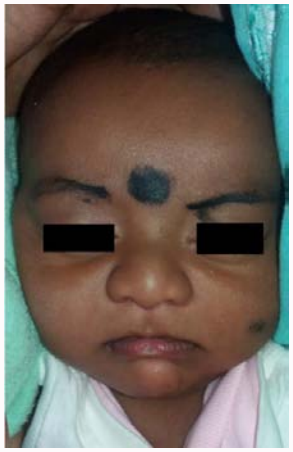

Figure 1: Extra oral image.

Figure 2: Polydactyly right hand (Mesio axial).

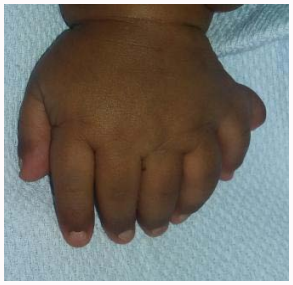

Figure 3: Polydactyly left hand (post axial).

in these genes, so, likely, other unknown genes are also responsible for EVC [1]. Not more than 25 cases have been reported in India. Around 150 to 170 cases were reported in the literature [5].

The syndrome occurs in many ethnic groups throughout the world and affects males and females in equal numbers. This condition has been reported in approximately 150 individuals. It is more common in the Old Order Amish population of Lancaster County, Pennsylvania, and in the native population of Western Australia.

Facies

The facies not especially characteristic, except for a mild defect in the middle of the upper lip. Usually, it is not a striking feature.

Skeletal anomalies

The extremities are often markedly shortened progressively distal wards, that is from trunk to phalanges. Bilateral postaxial hexadactyly is frequent and heptadactylus also can be noted. The patient can make

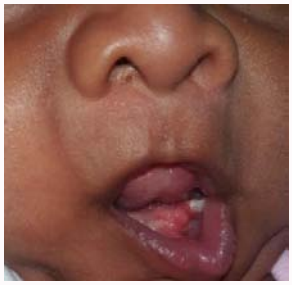

Figure 4: Upper lip with midline cleft, Natal tooth and bulbous tipped nose.

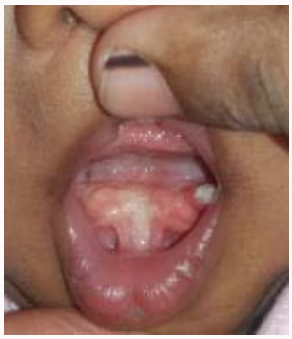

Figure 5: Multiple labial frenula of mandible.

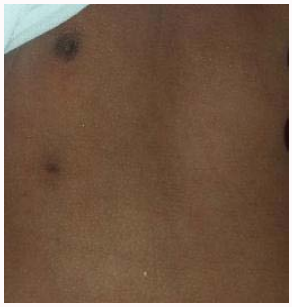

Figure 6: Accessory nipple in right chest.

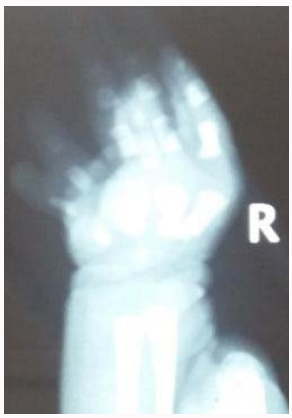

Figure 7: Right hand radiograph.

a tight fist often. Wide space may present between the hallux and other toes. Genu valgum, the curvature of humerus, pectus carinatum with thoracic constriction also noted.

Radiographically, the tubular bones are short and thickened. The diaphyseal ends of the humerus and femur are plump. The shortening

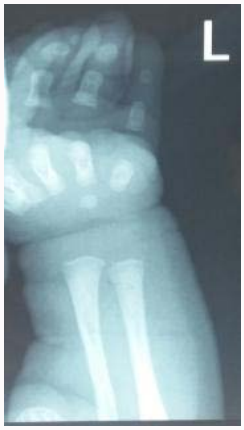

Figure 8: Left hand radiograph.

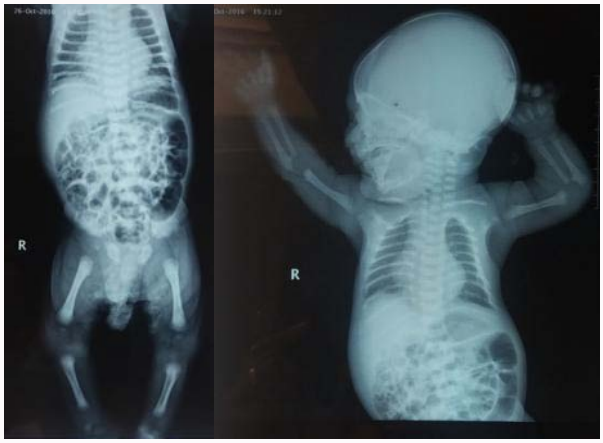

Figure 9, 10: Radiograph showing pectus carinatum, genua vulga and thoracic constriction.

of radius and ulna is even more marked than that of the humerus. The proximal ends of the ulna and distal end of the radius are unusually large, and the proximal end of the radius and the distal end of the ulna are unusually small. Syncarpalism, syn-metacarpalism, and polymetacarpalism are frequent. Cone-shaped epiphysis of hands is pathognomonic of this syndrome. In infancy, the pelvis is dysplastic with low iliac wings and hook-like downward projection of the medial acetabulum. In childhood, the pelvic shape normalizes.

Heart

Congenital heart defects are found in 50% to 60% [5].

Hair and nails

The hair particularly, the eyebrows and pubic hair are thin and sparse. Nearly all the patients have severe dystrophy of fingernails which are markedly hypoplastic, thin often wrinkled/spoon-shaped.

Eyes

Often normal but esotropia and congenital cataract have been observed.

Genitourinary system

About one-third of male patients have genital anomalies like cryptorchidism, hypospadias, etc.

CNS

Retardation is not an integral part of this disorder.

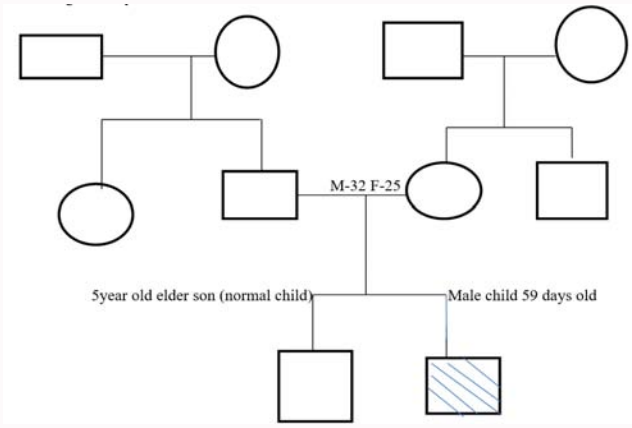

Figure 11: Pedigree analysis.

Oral manifestation

The most striking and constant finding is the fusion of the middle portion of the upper lip to the maxillary gingival margin so that no mucobuccal fold or sulcus present anteriorly. The middle portion of the upper lip appears to have a notch. Natal teeth are seen in at least 25% at least. Congenitally missing teeth also, a common finding particularly in the mandibular anterior region. Alveolar ridge often serrated in that region. Erupted teeth are usually small, conical-shaped. Supernumerary teeth were also reported [1].

Diagnosis

The diagnosis can be made as early as the 18th week of gestation by ultrasonography when the increased nuchal translucency is evident [6-8]. Ellis-Van-Creveld syndrome is diagnosed by the observation of short stature, slow growth, skeletal abnormalities determined by imaging techniques, and sometimes teeth present at birth (natal teeth). Molecular genetic testing for the EVC1 and EVC2 genes is available on a research basis only.

Differential diagnosis

Features in hands, pelvis, and long bones are similar to EVC in Thoracic dystrophy also. Differential diagnosis between both is based on cardiac anomalies, nail hypoplasia, the fusion of upper gingiva if present, neonatal teeth. Later in life, Genu valgum in EVC and renal failure with thoracic dystrophy distinguish two disorders. Other chondrodysplasias like Achondroplasia, Morquio syndrome are distinguished from EVC by radiographic features. Partial fusion of the upper lip as a result of the hyperplastic frenum is seen in orofacial digital syndrome. Natal teeth are observed in Pachyonychia congenita and Hallermann-Streiff syndrome [1].

Treatment

It is often necessary to treat respiratory distress shortly after birth that results from a narrow chest and/or heart failure. Natal teeth should be removed because they can interfere with feeding. The treatment of Ellis-Van Creveld syndrome is directed toward the specific symptoms that are apparent in each individual. Such treatment may require a multidisciplinary approach. Genetic counseling is recommended for affected individuals and their families.

Conclusion

A case has been reported here with classic features of Ellis-van Creveld syndrome. It is a rare congenital disorder, with high mortality in early life, 1/3 of these patients die in infancy from

cardiac and respiratory problems and those who survive to require a multidisciplinary approach for treatment.

Acknowledgment

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