

Chondroectodermal Dysplasia (Ellis-Van Creveld Syndrome): A case report

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Citation

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Abstract

Ellis –van Creveld syndrome is a rare autosomal recessive syndrome characterised by several skeletal anomalies, dental anomalies, nail dysplasia, oral mucosal anomalies and congenital cardiac defects. The authors here are reporting a case of Ellis-van Creveld syndrome in a thirteen month old female child presented with history of recurrent lower respiratory tract infections and was diagnosed incidentally as a case of Ellis –van Creveld syndrome

CASE REPORT

A thirteen months old female child presented with cough, breathlessness and fever . She was third in birth order, born of first degree consanguineous marriage. There was no history of infection, drug intake or radiological exposure during pregnancy. Family history was normal.

On clinical examination the patient had disproportionate short stature, the upper segment to lower segment ratio was 2:1 The height of child was 70cms against 85 cms of expected. Weight was 7 kgs against 9kgs of expected. Sparse hair and eyebrows with mild bifid philtrum was seen. Both hands had polydactyly on ulnar side and the nail hypoplasia was present (Fig.1)

Figure 1

Figure 1: showing nail hypoplasia and ulnar polydactyly



Fusion of upper lip and gingiva obliterating the sublabial sulcus and mandibular arch are showing multiple clefts (Fig 2 & 3).

Figure 2

Figure 2: showing fusion of upper lip with gingival sulcus



Figure 3

Figure 3: showing multiple clefts in mandibular arch



Peripheral pulses were normal in rate character and rhythm with no radiofemoral delay. There was mild pallor, no edema and lymph nodes were not palpable. Bilateral rales were heard in Chest on auscultation. Cardiac auscultation revealed a systolic murmur with a loud P2. Muscle mass and tone and reflexes were normal.

Figure 4

Figure 4: showing long narrow chest, cardiomegaly and curved humerus



Figure 5

Figure 5: showing unusually enlarged proximal ulna and distal radii with cone shaped epiphysis and the polydactyly



Radiographic evaluation of the chest and limbs revealed narrow chest with short ribs cardiomegaly and curved humerus(Fig 4).

X-ray of hands and distal upper limbs showed unusual enlargement of proximal ulna and distal radii, and unusually small proximal radii and distal ulna with cone shaped epiphysis of hand bones(Fig. 5). Radiographs of pelvis and thigh showed small iliac wings with hooked acetabulum and thick femur(Fig.6). X-ray of lower limbs showed hypoplastic tibial epiphysis with proximal tibial metaphyseal enlargement. (Fig.7)

Figure 6

Figure 6: showing small iliac wings with hypoplastic epiphysis of femur

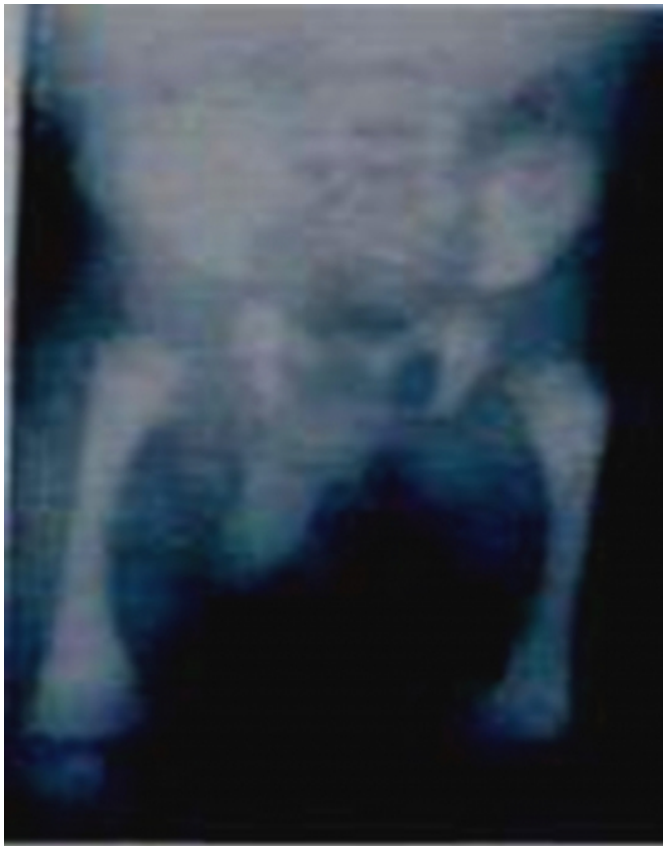
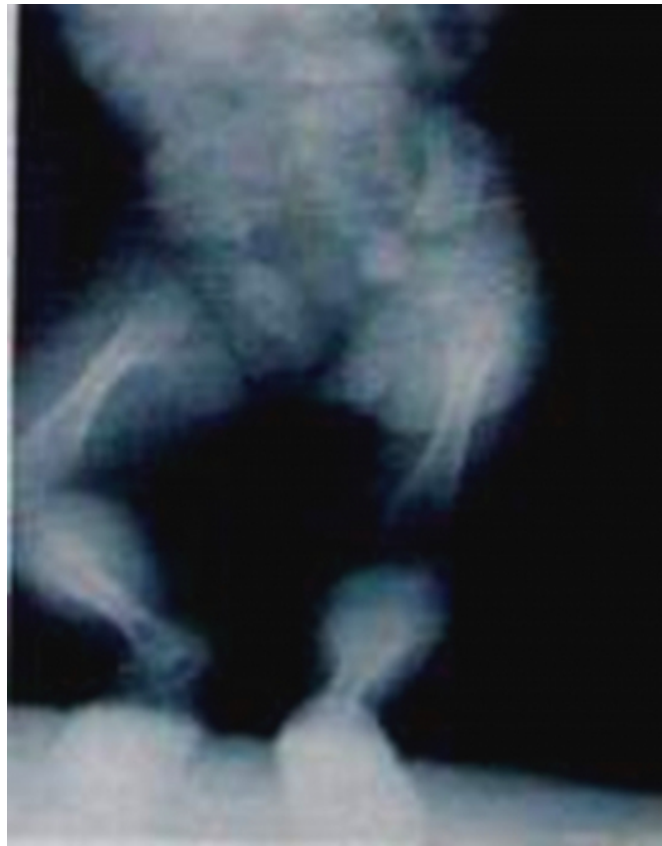


Figure 7

Figure 7: showing proximal tibial metaphyseal enlargement with short fibulas and hypoplastic epiphysis



Laboratory tests revealed a Hb of 8gms/dl, ESR of 35mm/hr (wintrob's method). ECG showed right axis deviation. On echocardiography, a large ASD signifying almost a single atrium with associated pulmonary arterial hypertension. Ultrasonography of the abdomen and cranium was normal. Karyotyping showed 46xx pattern. Metabolic screening was normal. Based on above clinical and radiological features a diagnosis of Ellis-van Creveld syndrome was made. The child responded to antibiotic and decongestive therapy and is doing well on follow up.

DISCUSSION

Chondroectodermal dysplasia or mesoectodermal dysplasia is a rare mesenchymal –ectodermal dysplasia and was first described in 1940 by Richard W.B. Ellis and Simon Van Creveld¹. This autosomal recessive condition is now usually called as Ellis- van Creveld syndrome and is usually seen in Amish community of Lancaster county Pennsylvania USA. Ellis- van Creveld syndrome is most common type of dwarfism among Amish community of Lancaster county Pennsylvania USA, occurring in 1/5000 live births and incidence in non Amish population is estimated as

7/1000000 live births₂

Ellis van Creveld syndrome comprises bilateral postaxial polydactyl, achondroplasia characterized by short limbs and ectodermal dysplasia. Congenital heart defects are also common.₃

Etiology is still speculative. However, recent identification of EVC gene has led to a better understanding of this syndrome. The EVC gene has been mapped to chromosome band 4p16 using linkage analysis. EVC2, that gives rise to the same phenotype of the syndrome.₄ These two genes lie in a head-to-head configuration that is conserved from fish to man.₅

The extremities are often plump.₆ Polydactyly is consistent finding in hands, which is bilateral post axial and on ulnar side. In 10% of cases polydactyly of feet is present. A wide space is often present between hallux and other toes.₇ Syncarpalism between hamate and capitate, and the synmetacarpalism and poly metacarpalism are frequently present. Histopathology of fetuses with Ellis-van Creveld syndrome revealed that cartilage of long bones shows chondrocyte disorganization in the epiphyseal growth zone. Variable chondrocyte disorganization revealed in central physeal growth zone of vertebrae. Ectodermal dysplasia is present in 93% of the reported cases. Hair particularly eyebrows and pubic hair is sparse and thin. Nails are hypoplastic, dystrophic, thin, spoon shaped or sometimes completely absent._{8,9} Fusion of middle portion of upper lip to gingival margin eliminating maxillary vestibule is a consistent feature of this syndrome.₁ These patients may have lip tie.

Cardiac anomalies are reported in upto 50-60% of patients of which 40% have single atrium. Some patients have cor trioculare or even corbioculare.₁₀ About 30% of the patients die of cardiac or respiratory problems during infancy. It has been demonstrated that there is paucity of intra-hepatic bile ducts progressing to cirrhosis and subsequently requiring transplant and in one case dyserythropoiesis is reported which may be of a coincidental occurrence or may represent an unusual EVC syndrome association._{11,12} Genitourinary anomalies are found in 22% of cases and include hypospadias, epispadias, hypoplastic penis, vulvar atresia, focal tubulomedullary dilatation, agenesis of kidneys and megareters. Although most patients have normal intelligence, occasional CNS anomalies and hydrocephaly have been noted with the Dandy-walker malformation has been associated in some rare cases.₁₃

Differential diagnosis of Ellis-van Creveld syndrome include short rib polydactyly syndrome type I (Sadino Noonan), there are frequently associated intestinal and genital malformations. In type II (Majewski), there is usually preaxial polydactyly with orofacial clefts. Type III (Verma Naumov) is similar to type I but there is less malformations of inner organs. In type IV (Beemer Langer) there is usually an omphalocoel but polydactyly is rare.₃ Other differentials include achondroplasia, chondrodysplasia punctata, asphyxiating thoracic dystrophy and Morquio syndrome.

Jeune dystrophy, McKusick-Kaufman syndrome and Weyers syndrome are the postnatal differential diagnoses for Ellis creveld syndrome. McKusick-Kaufman and Ellis-Van Creveld Syndrome are both recessively inherited disorders, share postaxial polydactyly and congenital heart defect. The distinguishing characteristics osteochondrodysplasia and ectodermal anomalies are seen in Ellis-Van Creveld Syndrome and McKusick-Kaufman syndrome has hydro metropolpos. McKusick-Kaufman is caused by mutations in a gene on chromosome 20p12, encoding a protein similar to members of the chaperonin family whereas Ellis-Van Creveld Syndrome is caused by mutation in EVC gene.

Management of such patients demands multidisciplinary approach. Pediatric, cardiac, genetic, pulmonologic, urologic, physical and occupational therapeutic as well as psychologic and rehabilitative management is needed. Orthopedic care should be sought in combination with surgical, orthodontic-prosthetic regimen to correct craniofacial morphology and teeth defects would be required for satisfactory cosmetic results. Preventive resin restorations may address the problem of recurrent caries.

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