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# A rare case report of an Indian child with Ellis Van Creveld syndrome

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**Abstract:**
Ellis Van Creveld syndrome (EVC) is a rare genetic disorder with autosomal recessive inheritance, seen more amongst the Amish population of Pennsylvania in USA with an incidence of 1:2,44,000." Six fingered dwarfisms" was another name given to this syndrome. This syndrome presents with four clinical characteristic features of ectodermal dysplasia, congenital heart disease, bilateral postaxial polydactyly and nail or teeth hypoplasia. A typical case of a 13-month-old child is present in this article

**Introduction:**
EVC syndrome described as chondroectodermal dysplasia; a rare genetic disorder of skeletal dysplasia. It's a single gene disorder autosomal recessively inherited due to mutations in EVC 1 and 2 genes located on the same chromosome 4p16. In 1940, the first case was described by Richard W.B. Ellis of Edinburgh and Simon Van Creveld in Amsterdam[1.] It is an uncommon syndrome with a prevalence of seven per 1,000,000 cases[2]. There is an equal predilection in males and females. It is more seen in people who are Amish of Lancaster country, Pennsylvania and in the Western Australia. It presents characteristically with the tetrad of (a) Chondroectodermal dysplasia (disproportionate dwarfism with short limbs and longer trunk), (b) bilateral postaxial polydactyly, (c) nail hypoplasia, dystrophic nails, malformed teeth and (d) congenital cardiac malformations, most commonly present as single atrium followed by septal defects [3]

**Case report:**
A 13-month-old male child, first order born to a second-degree consanguineous marriage was brought to OPD, for evaluation for unprovoked generalized tonic-clonic seizures. Antenatal and birth history was uneventful. Family history was unremarkable. Child was immunized and all milestones were age appropriate.
On examination the child was well-built with subtle dysmorphic features of depressed nasal bridge, midfacial hypoplasia, absent upper central incisors, post axial polydactyly in both upper and lower limbs, short stubby fingers and toes and clinodactyly. (Figure 1(a, b ,c). Height of child was 72cms (-2SD and -1SD) with upper segment to lower segment ratio of 1:1.22 short stature (Height - 72cms (-2SD and -1SD) with upper segment to lower segment ratio of 1:1.22, meso-melic limb shortening. Skin, hair and nails appeared normal. Height of the child was 72cms (-2SD and -1SD) with upper segment to lower segment ratio of 1:1.22 with short forearms and legs. A detailed clinical examination revealed normal neurological examination, other systemic examination was also normal. Furthermore, investigations were carried out 2D Echo doppler revealed small fenestrated secundum atrial septal defect. EEG and MRI brain was normal. Xray findings described in Figure 2(a, b) suggestive of polydactyly with duplication of fifth metacarpal, Chest Xray appears normal, pelvis and acetabulum appear normal. Based on the clinical features along with 2D Echo doppler findings, child was diagnosed as Ellis Van Creveld syndrome.

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Figure 1 (a)

Figure 1(b)

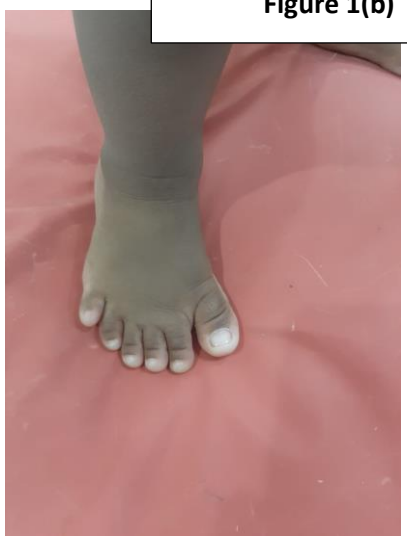

Figure 1 (c)

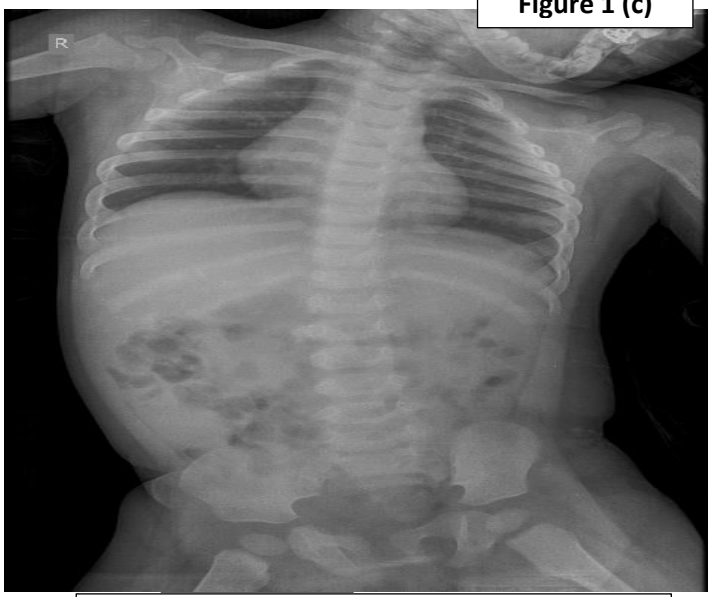

Figure 2 (a): Xray PA view appearing normal

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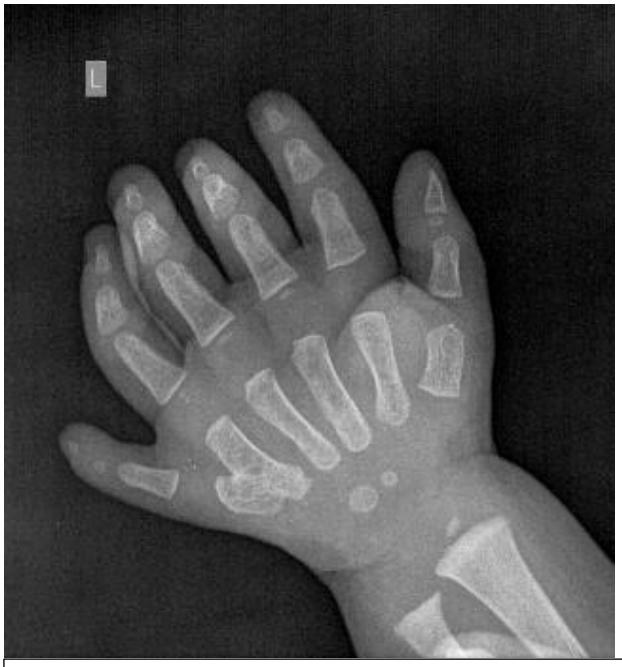


**Figure 2 (b):**

**XRAY left wrist joint PA and Lateral view showing Post axial polydactyly with duplication of fifth metacarpal.**

**Discussion:**

Ellis van crevald syndrome is a rare disorder which is inherited in an autosomal recessive manner. Although it is often found in Amish group, it is not reported to have a racial or gender predilection. It is also called as chondroectodermal dysplasia or meso-ectodermal dysplasia[3]. The characteristic feature of this syndrome is parental consanguinity or affected siblings Approximately 30% cases have parental consanguinity[4]. As observed by in a study siblings had a risk of 25% per offspring[5]. Our case also has history of second-degree parental consanguinity. EvC consists of following tetrad of features which are Bilateral manual postaxial polydactyly, long bone Chondrodysplasia resulting in Acro melic dwarfism, ectodermal dysplasia which has an effect on the nails, teeth, and hair, Congenital cardiac malformations: most often an atrial septal defect and endocardial fusion defect and hypoplasia of aorta[5.] Our case had an atrial septal defect.

Chondrodystrophy has been reported to be the most consistent finding, which is due to a defect in ossification resulting in short stature and limb shortening which is more distinct in the distal rather than proximal extremities[5.] Oral deformities such as a Partial harelip, maxillary alveolar clefts, abnormal tooth shape (conical teeth), deformities in size, structure, shape and number along with abnormal site of implantation is seen. In some cases, multiple labial frenum and obliteration of the labio-gingival sulcus have been seen, all considered pathognomonic and should be used in primary diagnosis[6].

Cardiac defects are found in 50–60% of patients with a common atrium and persistent atrioventricular canal being the most common defects. Patent ductus arteriosus, ventricular septal defects and atrial septal defects could also be present. This case had a n atrial septal defect. Radiological features comprise a narrow thoracic cage and wide, spade-like anterior ends. The pelvis has square iliac ala and "trident acetabulum." However, our case had a normal pelvis. Limbs have shortened long bones, with wide diaphysis and metaphysis. Proximal and middle phalanges are short with cone shaped epiphyses. Ellis–Van Creveld syndrome or chondroectodermal dysplasia has been mapped to chromosome 4p16[7.]

Kurian et al reported there were markedly hypoplastic nails, also with dystrophy and thinning, resulting them to take a spoon shape. Some instances also have found absence of nails. However, in our case they appeared normal.[8]

Additional clinical findings affecting other organs may occasionally be observed, although these were not diagnosed in our case.

Reports have mentioned Genitourinary anomalies presenting in 20% cases including agenesis and renal dysplasia, uretectasia and nephrocalcinosis[9]. Other infrequent features include strabismus, epispadias and hypospadias. Exceptionally, hematological anomalies have been reported.

Gokulraj et al, has reported Weyers acrofacial dysostosis, thoracic dysplasia of jeune, thoracic cage deformities, chondrodysplasia punctata and thoracic dystrophies to be a few close differentials of EvC syndrome.[6]

Some uncommon findings seen in EVC include Dandy Walker malformation and congenital cataracts.

Early diagnosis can be made antenatally at 18$^{th}$ week of gestation by ultrasonogram by measuring any significant increase in nuchal translucency. Findings may later on be confirmed by clinical examination[10]. The definitive diagnosis is genetic based where homozygosity for a mutation in the EVC1 and or EVC2 genes , gene is identified on short arm of chromosome 4 with five different mutations[11]. It is detected by direct sequencing. However, the gene mutation is positive in only two third of the patients.

An integrative approach to managing cases of EvC syndrome includes a multidisciplinary team of specialists.

The prognosis depends largely on the respiratory problems as a result of thoracic cage abnormality and degree of cardiac defects, those who survive infancy have a normal life expectancy, oldest living patient was 82 years of age[12.]

**Conclusion**

This case enlightens the insight into EvC syndrome, and will help readers gain idea about findings which are rare and help medical professionals diagnose, prognose and treat patients effectively and timely.

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