ELLIS VAN CREVELD SYNDROME

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Introduction

Chondroectodermal dysplasia is a complex genetic syndrome first described in 1940 by Ellis and Van Creveld (1940) which is also called mesoectodermal dysplasia, Ellis-Van creveld syndrome or chondro-dystrophy syndrome. It is a rare autosomal recessive disorder whose minimal diagnostic criteria include postaxial polydactyly of the hands, short limb dwarfism and dysplastic finger nails and teeth. We describe a case with shortening of limbs and polydactyly.

Case Report

A 7 months old female child was referred to our clinic for skeletal survey for clinically suspected achondroplasia. On clinical examination, the upper and lower limbs were shortened disproportionate to the trunk and polydactyly was noted in both hands and right foot.

Readiographic features

There is disproportionate mesomelic limb shortening with normal length of trunk. (Fig. 1)

The long bones are short and broad with bowing and dumb-bell appearances on both sides. The metaphyses are expanded and dumb-bell shaped most marked in proximal tibia. (Fig. 2)

There is disproportionate shortening of fibulae on both sides. (Fig. 2)

There is accelerated ossification of the proximal femoral capital epiphyses and proximal epiphyses of humeri. (Fig. 1 and 2)

The clavicles are placed upwards but otherwise normal. The ribs are short and thick. (Fig. 1)

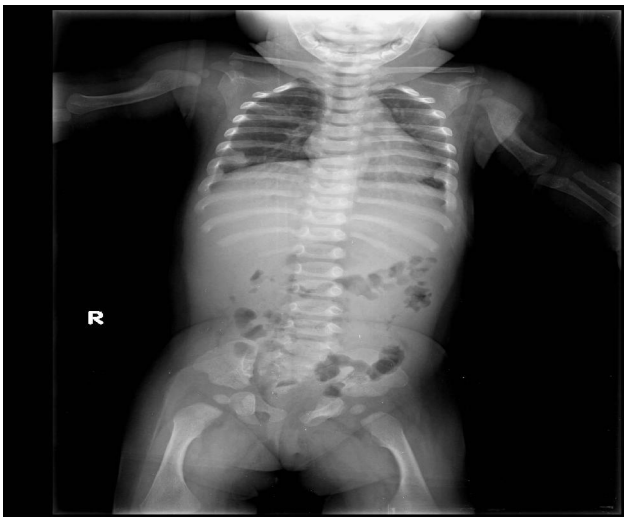

Figure 1

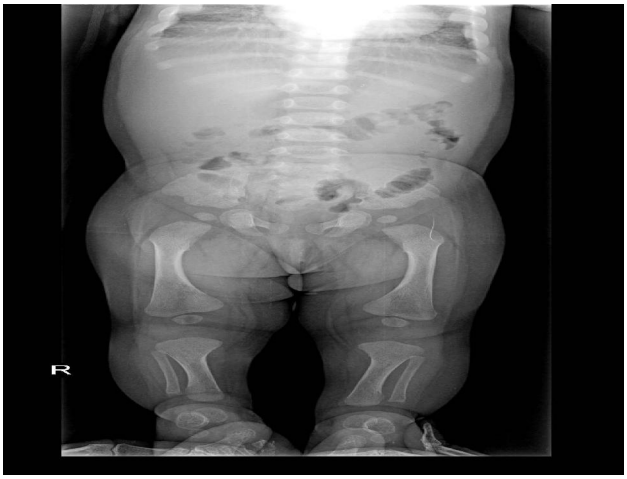

Figure 2

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There is polysyndactyly in both hands and right foot. There is evidence of absence of distal phalanges in all fingers and toes and absence of middle phalanges in the duplicated right 2nd toe and left 5th toe. Soft tissue swelling and thickening over the limbs seen. (Fig. 3, 4, 5, 6, 10)

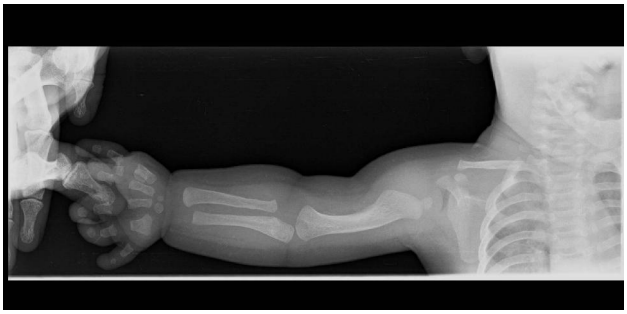

Figure 3

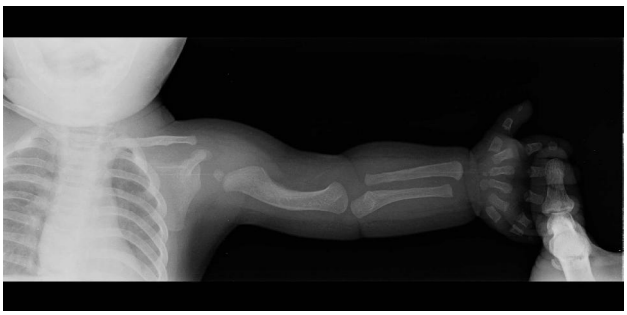

Figure 4

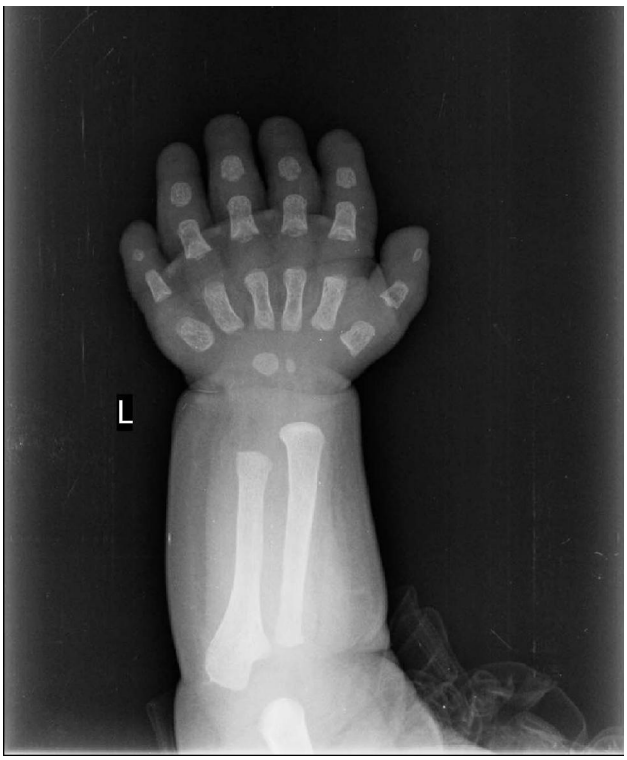

Figure 5

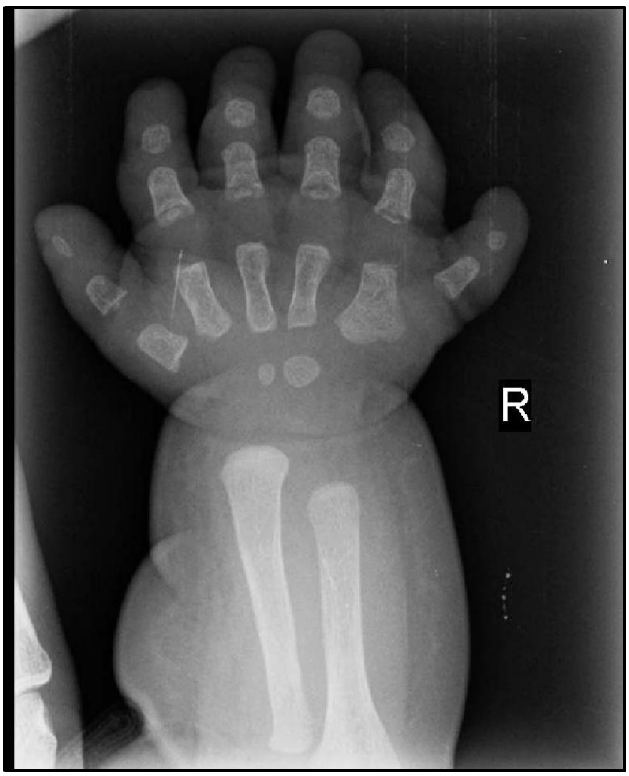

Figure 6

The iliac bones are short and square with transverse orientation of acetabulae, more marked on the left. (Fig. 2)

Multiple teeth seen in upper and lower jaws, wormian bones are seen in right coronal and lambdoid sutures. (Fig. 7, 8)

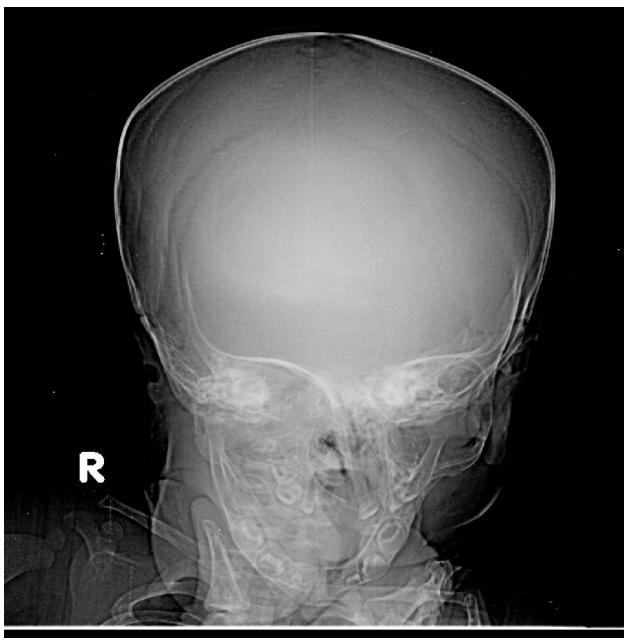

Figure 7

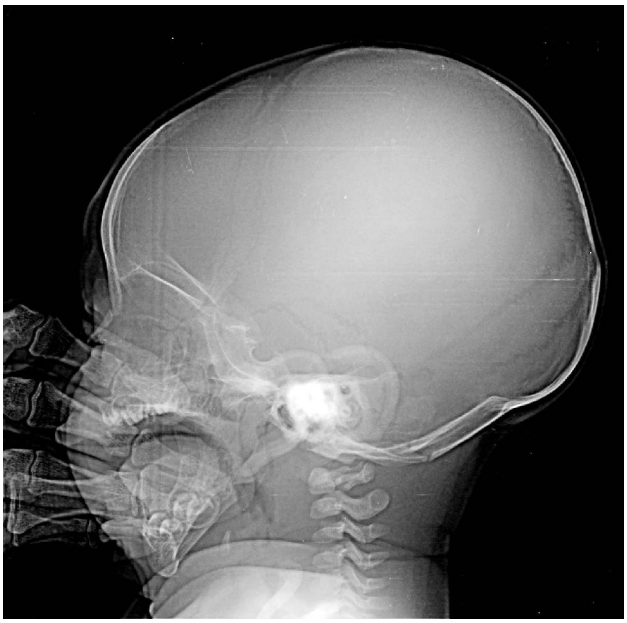

Figure 8

Spine is normal, no anterior or posterior scalloping seen. The interpedicular distances increase normally from L1 to L5 level. (Fig. 9)

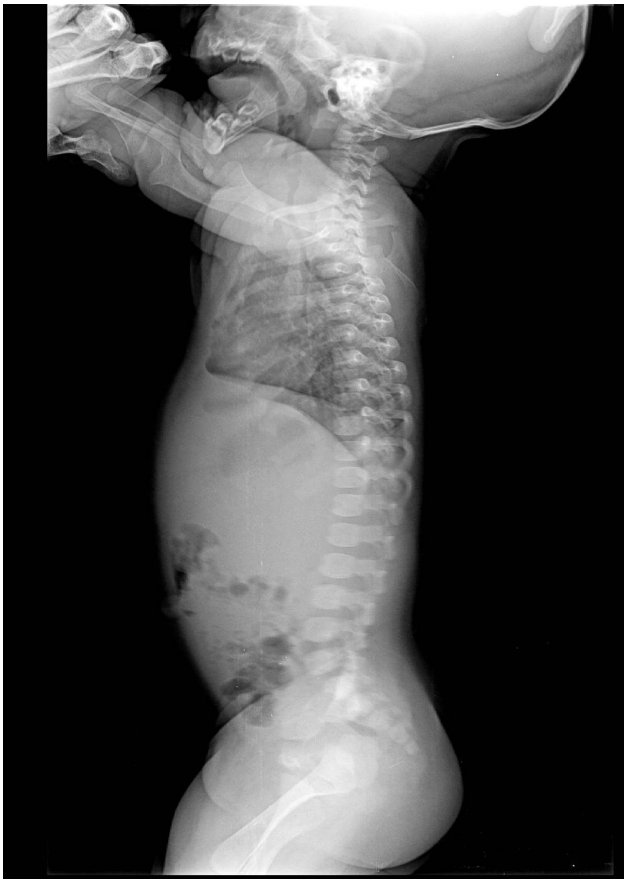

Figure 9

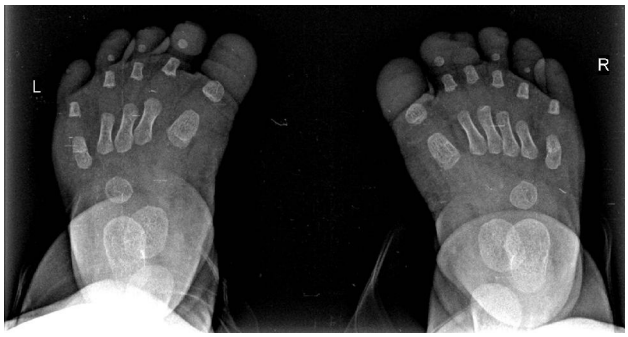

Figure 10

Discussion

Chondroectodermal dysplasia or Ellis-Van Creveld syndrome is rare genetic disorder. It belongs to the short rib dysplasia group of the osteochondrodysplasia and is inherited on an autosomal recessive basis, with considerable variability of expression. Some patient may reach adult height whereas in others a short stature persists. Its prevalence has been estimated at 0.9 per 100,000 births.

Ellis-Van Creveld syndrome presents phenotypic variation. Isolated findings in near relatives, such as polydactyly and short stature can be seen. A female sibling of our patient showed polydactyly and short stature. (Fig. 10)

Clinically it is characterized by a disproportionate short-limb dwarfism (with centrifugal shortening), postaxial polydactyly (hexadactyly) of the fingers and sometimes the toes hypoplastic nails, dental anomalies, multiple labiogingival frenula, cardiac abnormalities in upto 50% of patients and knock-knee deformity. The chest is usually short. Cardiac evaluation is the most important in the management of these patients with EVC as that determines survival and longevity. Our patient has no major or minor cardiac abnormality but chest is short. On complementary echocardiogram, no cardiac abnormality was found.

Major radiographic findings include short ribs, short and widened tubular bones, a small pelvis with short flared iliac bones and a flat acetabular roof with “three downward-projecting spikes” premature ossification of capital femoral epiphyses, polydactyly and cone-shaped epiphyses of the phalanges of

the hand. There was also sloping humeri which is typically only seen in EVC. Our patient has premature ossification of capital femoral and humeral epiphyses on both sides and polydactyly and short ribs.

In later infancy and childhood, fusion or over lapping of the capitate and hamate bones and hypoplasia of the lateral proximal tibial ossification centers with exostosis formation at the medial aspect of the proximal tibial shafts may occur.

Conclusion

We report a case of rare skeletal dysplasia. Her elder female sibling is also affected with same dysplasia while asymptomatic till date. Echocardiogram of the patient was also done to rule out any cardiac abnormality, the echocardiogram was unremarkable.

References

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