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CASE REPORT: RARE CASE OF MULTIPLE EPIPHYSEAL DYSPLASIA IN 2 YEAR OLD

JANANI S M

Second year Junior Resident, Department of Pediatrics, Sree Balaji Medical College and Hospital, Chennai

*Corresponding Author: Dr. Janani SM: Email: janniammu@gmail.com

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ABSTRACT

Multiple epiphyseal dysplasia is a group of disorder of cartilage and bone development, primarily affecting the ends of the long bones in the arms and legs (epiphyses) **[3]**. Two types of MED which are distinguished by their patterns of inheritance-autosomal dominant and autosomal recessive characterised by short stature, joint pain, waddling gait, joint deformity (valgus) , early onset osteoarthritis with normal mental development and blood parameters.

CASE

A2 year old girl presented to pediatrics outpatient clinic with abnormal positioning of left leg while walking.

On examination: she noted to have short stature (<-3SD) with dolichocephaly , frontal bossing, protruding abdomen , genu valgum deformity of left leg and vertical talus **[2]**. Normal gross and fine motor development and normal mentation. Family history of short stature present. Paternal side skipping of generation is seen. Vitamin D, Phosphorous, Calcium, thyroid profile are within in limits.

Xray of hand showed short metacarpal, lower limb xray showed Genu valgus deformity of leg with flattened epiphysis of upper end of left femoral head and flattening epiphysis of lower end of tibia and feet showed equinus deformity **(Figure 1, 2) [1-4]**.

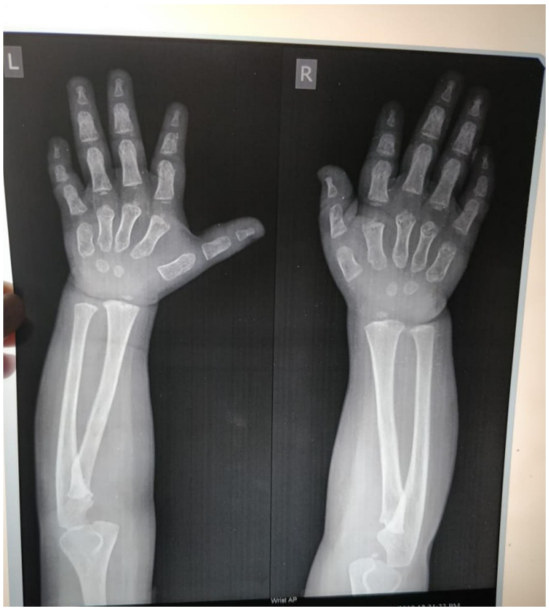

Figure 1

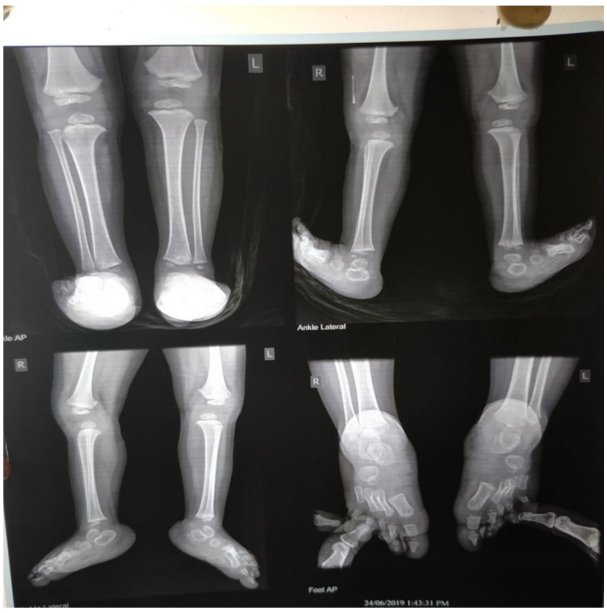

Figure 2

DISCUSSION

Flattening of the epiphyseal of long bones is hall mark of multiple epiphyseal dysplasia. The deformity persist throughout life and cause premature osteoarthritis **[1]**. There is no characteristic changes in the metaphysis. Differential diagnosis of the above case which needs to be considered are spondyloepiphyseal dysplasia, pseudo-chondroplasia and perthes disease. There was no platyspondyly in this case and the trunk was not disproportionately short so spondyloepiphyseal dysplasia ruled out. In pseudoachondroplasia there are characteristic vertebral changes, flared

metaphysis, small irregular epiphysis which were not present in this case. Features of perthesdisese like erosion of involved epiphysis with irregular flattened and fragmented epiphysis were not seen.

CONCLUSION

Multiple epiphyseal dysplasia is one of the rare cause of short stature. It should be considered when there is family history of short stature and all other causes of short stature ruled out. Diagnosis is based upon identification of bony abnormalities, a detailed family history and characteristic X ray features **[4]**.

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