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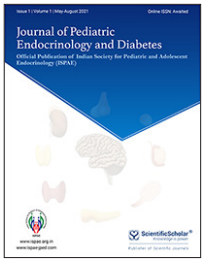

Clinical Images/Spotters

Metaphyseal dysplasia – An uncommon mimicker of rickets

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ABSTRACT

Metaphyseal dysplasias are rare conditions of genetic origin with clinical features similar to rickets. Awareness of these conditions is warranted to avoid delay in diagnosis. It will also help avoid unnecessary treatment with vitamin D leading to toxicity. We report here a case of a toddler who presented with clinical and radiographic features of rickets, who was eventually identified as a case of autosomal recessive metaphyseal dysplasia, Spahr type (OMIM# 250400).

Keywords: Metaphyseal dysplasia, *MMP13* gene, Vitamin D deficiency rickets, Metaphyseal anadysplasia, Metaphyseal chondrodysplasia Spahr type

CASE SUMMARY

A 10-month-old male infant was incidentally noted to have clinical features suggestive of rickets while hospitalized for bronchopneumonia. He was the first-born child of unrelated parents and was healthy, and thriving with normal developmental milestones attained to date. His weight was 8.1 kg (World Health Organization [WHO] Z-score –0.85), length was 75 cm (WHO Z-score +0.90), and head circumference was 45.5 cm (WHO Z-score: +0.41). He had wide wrists [Figure 1] and Harrison's sulcus. His anterior fontanel was closed. The spine, hair, teeth, and nails were normal. The serum calcium was 9.9 mg/dL (normal range 8.8–10.8 mg/dL), phosphorus 6.5 mg/dL (normal range 3.7–5.6 mg/dL), alkaline phosphatase (ALP) 360 IU/mL (normal range 145–420 IU/L), and 25-hydroxyvitamin-D3 19 ng/mL (normal range 30–70 ng/mL). The X-ray of his wrists showed irregular widened metaphyses [Figure 2]. X-rays of the spine were unremarkable with no radiological evidence of osteopenia. He received therapeutic doses of vitamin D followed by prophylaxis to correct low vitamin D3 levels. On follow-up, clinical and radiological features of rickets persisted, without any biochemical evidence of rickets. This led to the consideration of a rickets mimicker like metaphyseal dysplasia. Parents were counseled and with their consent, genetic studies (exome sequencing by next-generation sequencing) were performed which detected compound heterozygous mutation in the *MMP13* gene. Sanger sequencing of the identified variants in the parents suggested the possibility of autosomal recessive metaphyseal dysplasia Spahr type (MDST) in our patient.

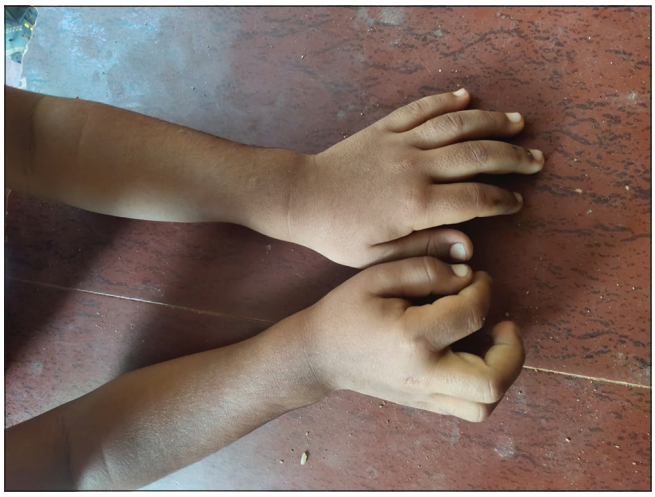

Figure 1 : Clinical photograph showing wide wrists.

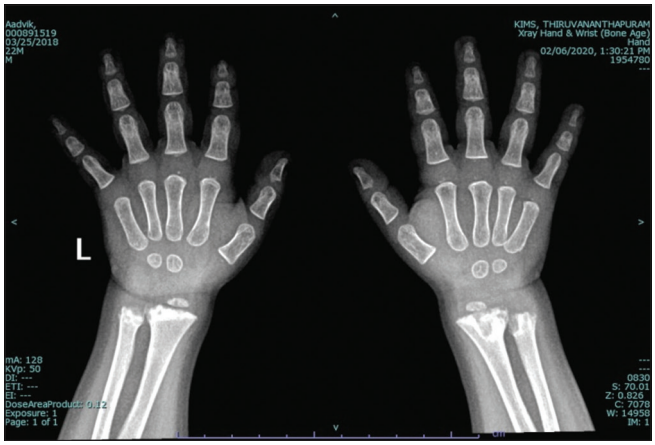

Figure 2: The X-ray of wrists showing irregular widened metaphyses.

DISCUSSION

Clinical and radiological features of rickets with normal serum ALP and with poor response following vitamin D therapy should alert a physician to the possibility of a rickets mimicker, the most common of which is skeletal dysplasia. The three forms of metaphyseal chondrodysplasias with overlapping clinical features are MDST, metaphyseal chondrodysplasia Schmid type, and metaphyseal anadysplasia.[1,2]

MDST is a heterogeneous group of disorders caused by a recessive mutation in the *MMP13* gene on chromosome 11q22.[3] The condition presents with abnormalities in skeletal growth and radiological features of rickets in the form of metaphyseal irregularities. Mild bone fragility may be a feature.

The early presentation in our child suggested the possibility of metaphyseal anadysplasia. However, clinical exome studies pointed to a diagnosis of MDST. The two compound heterozygous variants identified and reported in the index patient lie in the catalytic domain of the MMP13 protein. The genotype-phenotype correlation, parental segregation studies, and prediction data strongly suggest that the variant is pathogenic in this child.

Declaration of patient consent

Patient's consent not required as patients identity is not disclosed or compromised.

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Nil.

Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-Assisted technology for manuscript preparation

The author(s) confirms that there was no use of Artificial Intelligence (AI)-Assisted Technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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