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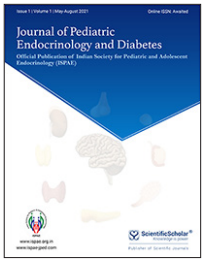

Clinical Images

Short child with hypermobility of shoulder joint: A rare case of skeletal dysplasia

T. N. Keerthana, H. N. Vani, Palany Raghupathy

Department of Pediatric Endocrinology, Indira Gandhi Institute of Health Sciences, Bengaluru, Karnataka, India.

***Corresponding author:**
T. N. Keerthana,
Department of Pediatric Endocrinology, Indira Gandhi Institute of Health Sciences, Bengaluru, Karnataka, India.

keerthana4tn@gmail.com

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A 6 year and 8-month-old boy, born of non-consanguineous marriage, presented with short stature from infancy and abnormal gait noticed from the age of 3 years. His weight was 14.6 kg (−3.11 SD), head circumference 50 cm, and height 101.3 cm (−2.5 SD). The height was within the mid-parental target height range (MPH at −6.6 SD) and upper segment to the lower segment ratio was 1.18:1. On examination, he had a wide-open anterior fontanelle (AF), unfused metopic suture, frontal and parietal bossing, normal dentition without any supernumerary teeth, hypertelorism, hypermobility of shoulder joint [Figure 1], and waddling gait. The mother and maternal grandfather had similar phenotype. Mother's height was 140 cm (−3.3 SD). Radiological evaluation showed wide open AF

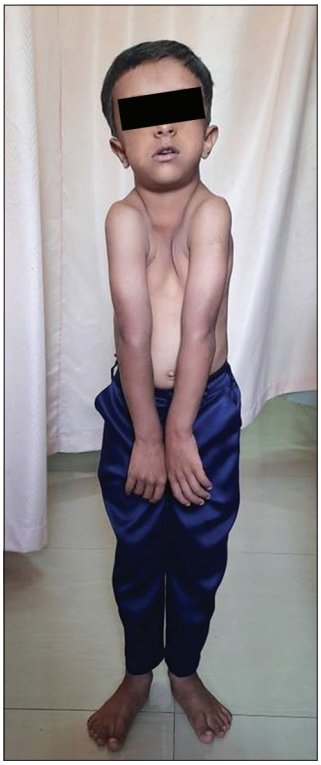

Figure 1: Hypermobility of shoulder joint.

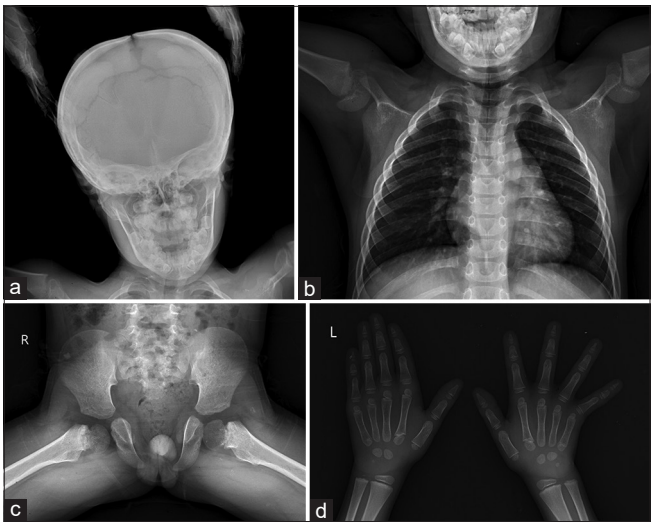

Figure 2: (a) X-ray skull showing wide open AF, (b) X-ray chest showing absent right clavicle and hypoplastic left clavicle, (c) X-ray pelvis showing shallow acetabulum, dislocated femoral head and pseudoarthrosis, (d) X-ray hand showing accessory epiphyses in 3rd and 5th metacarpals bilaterally. AF: Anterior fontanelle.

[Figure 2a], wormian bones around lambdoid suture; absent right clavicle, hypoplastic left clavicle [Figure 2b]; shallow acetabulum with dislocated femoral head and pseudoarthrosis [Figure 2c]; and accessory epiphyses in 3rd and 5th metacarpals bilaterally [Figure 2d]. Mother had similar phenotype with wide open AF with unfused metopic suture [Figure 3a] and radiological features of bilateral hypoplastic clavicle [Figure 3b] and wide open AF [Figure 3c]. Thus, the child was diagnosed to have cleidocranial dysostosis.

Cleidocranial dysostosis is a rare skeletal dysplasia with prevalence of 1 in 1,000,000, with autosomal dominant inheritance.[1] The gene responsible for this condition, *RUNX2* is located on chromosome 6p. Cleidocranial dysostosis has clinical triad of delayed closure of cranial sutures, hypoplastic clavicle, and dental abnormalities.[2] Management includes using safety helmets to avoid head trauma and early dental evaluation. There is no role of growth hormone therapy. Prenatal genetic counseling is helpful. All children with disproportionate short stature need detailed clinical and radiological evaluation to rule out skeletal dysplasias, before screening for growth hormone deficiency.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

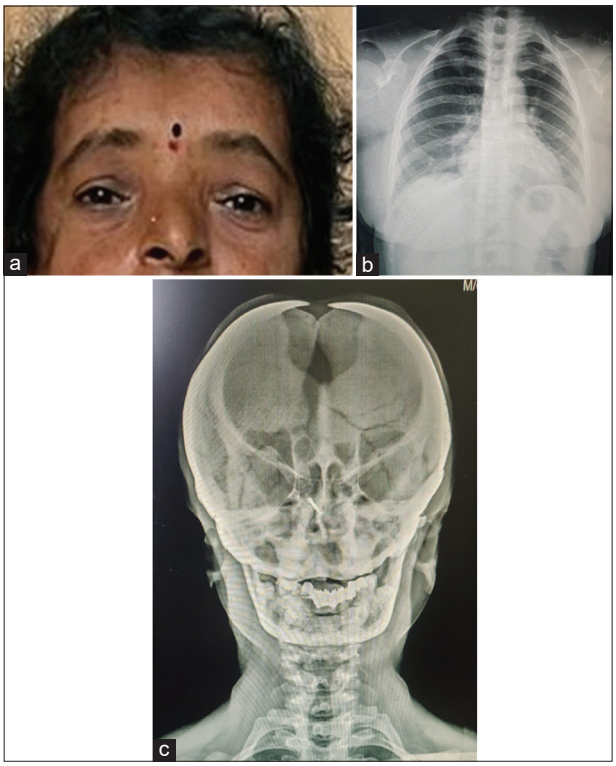

Figure 3: (a) Mother has depression of forehead in midline due to delayed fusion of metopic suture; hypertelorism, (b) mother's chest X-ray showing bilateral hypoplastic clavicle, (c) mother's skull X-ray showing wide open AF. AF: Anterior fontanelle.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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