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Visual Diagnosis

Unilateral Bowing of Legs — Do not Forget to Examine the Skin

Arundhati Banerjee, DM, Swetlana Mukherjee, MD, Sweta Shah, MD, Lokesh Saini, DM *

Department of Pediatrics, Post Graduate Institute of Medical Education and Research, Chandigarh, India

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This five-year-old boy presented with nonprogressive unilateral segmental bowing of the lower part of left leg (Fig 1) noticed from early infancy. He had not experienced pain, swelling, or redness. There was no history of trauma. The child was born to nonconsanguineous parents with an uneventful perinatal period. His developmental milestones were age appropriate. On examination, he was a thriving child with normal anthropometric parameters. There was no evidence of micronutrient deficiency, wrist widening, rachitic rosary, Harrison groove or sulcus, frontal bossing, or recurrent chest infections in the past. His gait was normal.

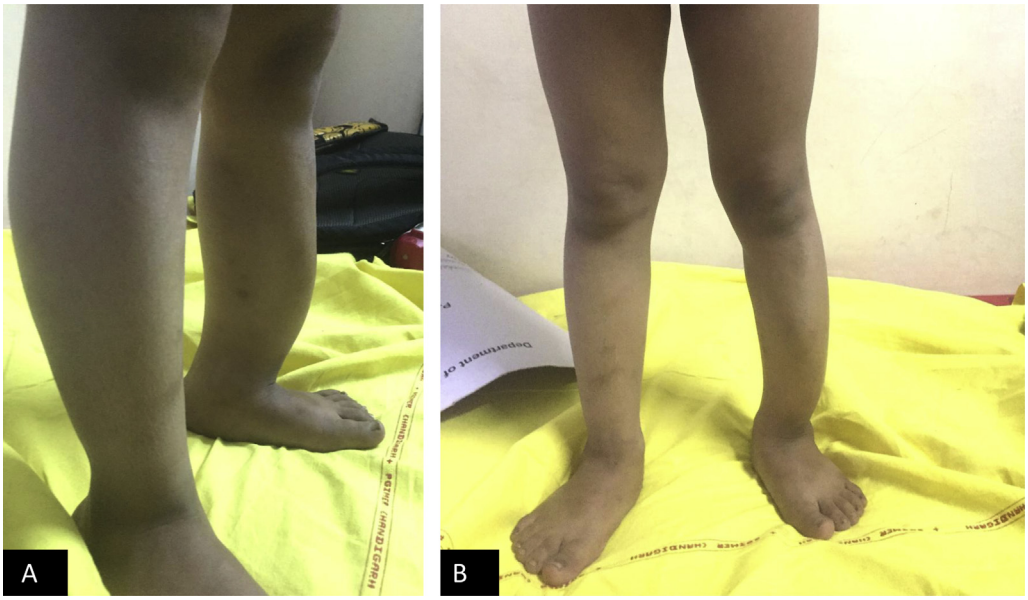

FIGURE 1. (A) Asymmetric anterior bowing of the left tibia. (B) Faintly evident bilateral hyperpigmented café au lait macules in both the legs, suggestive of neurofibromatosis. The color version of this figure is available in the online edition.

Conflicts of interest: None reported.

* Communications should be addressed to: Dr. Saini; Assistant Professor; Pediatric Neurology Unit; Department of Pediatrics; Post Graduate Institute of Medical Education and Research; Sector, 12; Chandigarh 160012, India.
E-mail address: drlokeshsaini@gmail.com (L. Saini).

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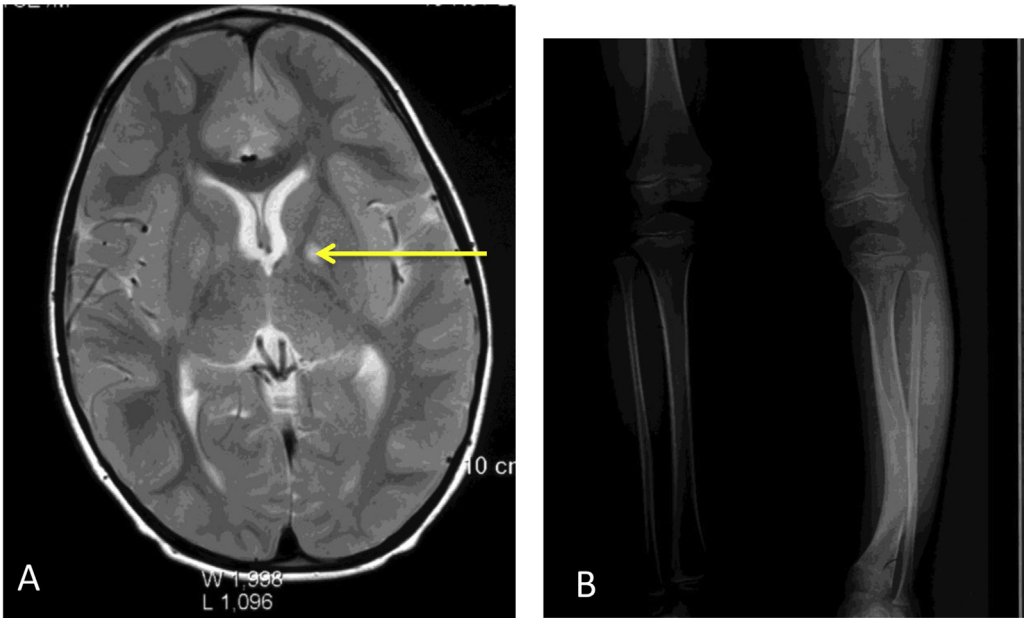

FIGURE 2. (A) MRI brain, axial T2 sequence at the level of the basal ganglia, revealed T2 focal hyperintensities (arrow) in the basal ganglia bilaterally (B) Skiagram of the legs (anteroposterior view) shows bowing of the tibia without rachitic cupping or fraying of metaphyses. MRI, magnetic resonance imaging. The color version of this figure is available in the online edition.

For this deformity, the child visited several practitioners who suspected it to be rickets its rickets, like hypophosphatemic rickets, due to selective lower limb involvement. He was prescribed multiple courses of vitamin D and calcium supplements without any improvement. On examination, he had multiple café au lait macules on the trunk and abdomen, previously unnoticed, leading to the diagnosis of neurofibromatosis. Skiagram of the left leg, anteroposterior view, showed bowing of left tibia without the classical deformities described in rickets like the widening of epiphyses and cupping and fraying of the metaphyseal end of the tibia. Magnetic resonance imaging of the brain (Fig 2) revealed multiple cortical, subcortical T2 hyperintense lesions qualifying as focal areas of signal intensities in bilateral basal ganglia, thalamus, cerebellum, and brainstem.

Discussion

Neurofibromatosis is a common neurocutaneous syndrome with an estimated prevalence of one in 4000.[1] Neurofibromatosis occurs due to dysregulation of the reticular activating system-mitogen activated protein kinase pathway, which is responsible for the disorder's varied manifestations.[2] Skeletal manifestations in neurofibromatosis are scoliosis, kyphosis, sphenoid dysplasias, nonossifying fibromas of the ends of long bones, and tibial bowing.[3] Dysregulated proliferation of osteoclasts relative to osteoblastic activity is responsible for the various bony deformities.[4] Tibial bowing is typically seen in the first year of life and involves the anterior shin bone; if not timely surgically managed it will lead to fracture of the midshaft with pseudoarthrosis formation and even limb amputation.[5] Such bowing often mimics rachitic conditions, but early onset, unilaterality, focal deformity, and careful clinical examination for café au lait macules should prevent unnecessary investigations in these children.

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