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Letter to Editor

Congenital Pseudoarthrosis Tibia with Fibrous Hamartoma in a Child with Neurofibromatosis

Sir,

Congenital pseudoarthrosis tibia (CPT) refers to the non-union of a tibial fracture that develops spontaneously after trivial trauma in a tibial diaphyseal segment. It usually develops during the first two years or sometimes later in life. The etiological nature of this lesion is unclear, however there is a strong association between CPT and neurofibromatosis type I (NF-I). Overall, 55% of the patients of CPT have evidence of NF-I, however only 6% patients with NF-I develop tibial dysplasia.[1] It is a rare condition with a reported incidence by Andersen of 1/190,000 live births.[2]

We report a case of a 5-year-old girl child who presented to the orthopaedics out patient department with a history suggestive of a left sided tibial fracture. She also had history of repeated tibial fractures since birth for which she was operated multiple times in private hospitals for which no records were available. On general physical examination, the child was pale, with presence of multiple cafe-au-lait spots on the back. On fundoscopic examination, Lisch nodules were seen. No neurofibromas/soft tissue nodules were observed. X-ray examination revealed a pseudo joint in left tibia with soft tissue swelling around the fracture site. Peroperatively, L shaped peudoarthrosis was resected and replaced by a fibular graft with K wire fixation done. The L shaped pseudoarthrosis with the fibrous cuff surrounding it was sent for histopathological examination [Figure 1]. Microscopic examination confirmed the presence of unremarkable bony trabeculae enclosing soft tissue comprising of bland spindle shaped cells [Figure 2].

CPT of long bones is a rare and complex orthopaedic condition characterised by failure of normal bone formation. Hamartomatous tissue forms at the site of fracture and pseudoarthrosis results because normal callus cannot form. Apart from tibia, it has been reported to occur in humerus, radius, ulna, clavicle.[3]

Neurofibromatosis type I is diagnosed in any patient who has two or more of the following seven signs:[4]

- Six or more cafe-au-lait macules >5 mm in pre-pubertal individuals or >15 mm after puberty
- Two or more neurofibromas of any type or one or more plexiform neurofibromas
- Axillary or inguinal freckling
- A tumor of the optic pathway
- Two or more Lisch nodules (iris hamartomas)
- A distinctive osseous lesion, such as sphenoid wing dysplasia or thinning of the cortex of the long bones (with or without pseudoarthrosis)
- A first degree relative with NF-I by above criteria.

Neurofibromatosis is one of the most common genetic

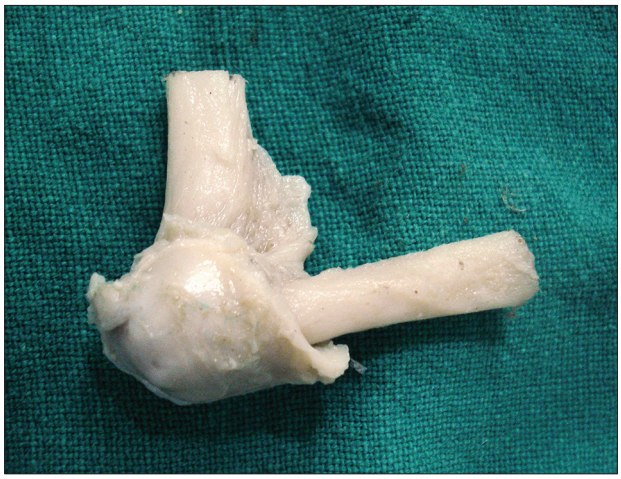

Figure 1: Gross photograph of the L shaped excision specimen of pseudoarthrosis, left tibia

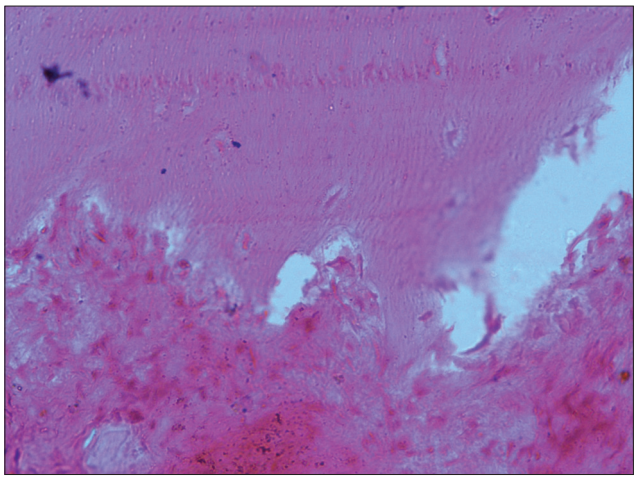

Figure 2: Tissue section from site of pseudoarthrosis showing mature lamellar bone along with soft tissue showing spindle cell proliferation: Fibrous hamartoma (H and E, ×400)

disorders seen by paediatric orthopaedic surgeons. Apart from CPT, other orthopaedic abnormalities in patients with NF-I include scoliosis, interosseous bone lesions, protrusion acetabuli and hemihypertrophy. Fibrous hamartoma is a key pathologic component of CPT, a challenging and disabling bone disorder. The main histopathologic change seen is the growth of a highly cellular fibromatosis like tissue which has been histochemically and immunohistochemically proven to be compatible with hamartoma.[5] Fibrous hamartoma cells maintain the mesenchymal lineage cell phenotypes but do not undergo osteoblastic differentiation in response to bone morphometric protein.

Congenital pseudoarthrosis tibia remains one of the most perplex and challenging orthopaedic disorder characterised by anterolateral deformity of tibia and shortening of the limb. The goal of the treatment is resection of the pseudoarthrosis and bridging the gap. The main problem in CPT is a biological one, where the osteogenic power at the pseudoarthrosis site is lacking, future researches should focus on finding out treatment modalities like bone morphometric protein,[6] that stimulate osteogenesis and bone repair.

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