



Case report

Congenital Pseudarthrosis of the Tibia: A Rare Orthopaedic Entity-A Focused Review

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Abstract

Congenital pseudarthrosis of the tibia (CPT) is a rare childhood disorder that may begin as anterolateral bowing and progress to established false-joint nonunion, refracture, limb-length discrepancy and disability. This focused review summarizes the clinical relevance, proposed pathophysiology, association with neurofibromatosis type 1, imaging findings, and treatment principles of CPT. Careful distinction between precursor bowing and established pseudarthrosis is emphasized, and an individualized multidisciplinary approach is recommended.

Introduction

The tibia is one of the bones of the lower limb and articulates with the fibula. Congenital pseudarthrosis of the tibia (CPT) is a rare but clinically important orthopaedic deformity of childhood; reported incidence varies between 1:140,000 and 1:250,000 [1]. Although usually unilateral, CPT is significant because the dysplastic segment may progress from anterolateral bowing to frank nonunion/pseudarthrosis, with recurrent fracture, limb-length discrepancy, mechanical-axis deviation and long-term disability [1-3]. A clear distinction is required between congenital anterolateral tibial bowing, which may be an early phenotypic pattern, and established pseudarthrosis, which implies a false joint/nonunion; bowing without nonunion should not automatically be labelled CPT. The fibula may be affected in about one third of cases, whereas bilateral occurrence is rare [4,5]. This focused review therefore emphasizes clinical relevance, proposed etiology/pathophysiology, NF1 association, imaging and treatment principles rather than an individual case narrative.

Paget first described congenital pseudarthrosis of the tibia in 1891. Pathologically, established CPT is characterized

as non-union of a tibial diaphyseal fracture that develops spontaneously or after trivial trauma in a previously dysplastic segment; refracture is common [1,2]. Anterolateral bowing, failure of the process of tubulation in early life, and cystic change before fracture are recognized precursor/risk patterns that may progress to deformity and sclerosis; however, these features alone do not establish pseudarthrosis until a false joint/nonunion is present [3,6].

In CPT, the tibia may show segmental dysplasia with anterolateral bowing. Osseous dysplasia can lead to tibial non-union; with distal tibial growth disturbance, this produces limb shortening. Early case and histopathologic studies proposed a dysplastic periosteal process with fibromatous or myofibroblastic overgrowth [7-9]. Later clinicopathologic observations support increased osteoclastic activity, altered signaling, reduced osteogenic potential, and impaired vascularization, accompanied by reactive medullary change, medullary sclerosis with excess trabecular deposition and, in some cases, intracortical cyst formation from excessive resorption [3,6]. Histopathologically, pseudoarthrotic tissue may contain fibrous tissue, fibrocartilage and hyaline cartilage with foci of enchondral ossification; spaces and clefts can be

lined by synovial-like tissue, and marrow spaces at the bone ends may be relatively devoid of haematopoiesis [3,6]. These observations support impaired bone formation and remodelling rather than a single uniform enzymatic cause.

Neurofibromatosis type 1 (NF1) is associated with CPT in a substantial subset of patients [10,11]. Sibling reports of congenital tibial defects with neurofibromatosis further support this link [12]. NF1 encodes neurofibromin, a negative regulator of RAS; loss of neurofibromin can maintain RAS-GTP signaling and disturb RAS-MAPK pathway regulation, with proposed effects on osteoblast differentiation and osteoclast activity. This mechanism is biologically plausible and may contribute to cyst formation and excessive bone resorption in NF1-associated CPT, but it should not be interpreted as the sole cause of all CPT cases. NF1 is an autosomal dominant disorder characterized by café-au-lait spots, neurofibromas, and, in some patients, pseudarthrosis. In selected CPT tissues, NF1 inactivation or “double activation” has been reported; in other cases, the diseased tibia shows deficient osteogenesis and mechanical strength with leg shortening [3,11]. Because NF1 is commonly mutated and pleiotropic, its presence should be documented clinically when suspected, while recognizing that CPT can occur without NF1 [10,13]. Abnormal fibrocellular and fibrovascular tissue at the cortex may promote osteoclastic resorption and abnormal modelling, producing fibrous hamartoma and pathological periosteum that interpose at the nonunion and impair vascularization.

Clinical and radiologic evaluation

The disease may become evident within the first few months of life or may be detected later; late-detected defects are usually more difficult to treat and, once bowing is severe or fracture has occurred, spontaneous healing is unlikely. Clinical assessment should document age, sex, family history/ NF1 features, limb-length discrepancy, mechanical axis, joint stiffness, and previous fractures or operations. Simple radiography clearly shows the defect; over the disease course, radiographs may show heterogeneous lesions, progressive deformity and non-union, with thin/atrophic or wide/hypertrophic tibial bone, often a cupped proximal and pointed distal fragment, and frequent fibular involvement. Bowing is easily seen radiographically, whereas MRI better defines bone and soft-tissue extent and is useful when planning treatment [2].

Management principles

Treatment is patient-specific and depends on bowing, limb-length discrepancy, bone quality, previous refractures, and NF1 status. The aims are durable tibiofibular union, correction or prevention of limb-length discrepancy, restoration of mechanical axis, and avoidance of soft-tissue injury, adjacent joint stiffness, and pathological fracture. Invasive treatment generally involves excision of diseased/pseudoarthrotic tissue followed by stable internal or external fixation with vascularized or nonvascularized graft; intramedullary nailing with bone graft, vascularized fibular transfer, and Ilizarov methods are established options [14]. Non-invasive or adjunctive

strategies, including electrical stimulation and replacement of diseased periosteum with iliac periosteal grafts, have been reported, but superiority of one method over another has not been definitively proven. Bone morphogenetic proteins have been used for malunion/nonunion and reported as adjuncts in children, including rhBMP-7 and rhBMP-2; however, evidence remains limited and use should be individualized [15,16] (Figures 1,2).



Figure 1: Radiograph Demonstrating Pseudoarthrosis of the Tibia



Figure 2: Radiograph Demonstrating Pseudoarthrosis of the Tibia.

Conclusion

CPT is rare but can be progressive and disabling because of pseudarthrosis, refracture, and limb shortening. This focused review summarizes clinical recognition, NF1 association, imaging evaluation, and contemporary treatment principles. When presented as a case report, age, sex, clinical history, examination findings, diagnostic work-up, treatment, follow-up, and outcome should be reported explicitly; such patient-specific data were not available in the material reviewed here, so no individual clinical course or outcome is claimed. For practice, early referral to a pediatric orthopaedic service, careful distinction of bowing from established pseudarthrosis,

NF1 assessment and individualized longitudinal management are essential to limit disability.

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