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REVIEW ARTICLE

Congenital pseudarthrosis of the tibia☆

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KEYWORDS
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Vascularized fibular grafts;
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Summary Congenital pseudarthrosis of the tibia (CPT) is an uncommon disease with various clinical presentations ranging from simple anterolateral tibial angulation to complete non-union with extensive bone defects. Classifications of radiographic findings include atrophic or hypertrophic pseudarthosis as well as cystic or dystrophic lesions. Although the relationship between CPT and type 1 neurofibromatosis is well known, the exact pathogenesis still remains unclear. The fibrous soft tissue found in the pseudarthosis and the abnormal periosteum are certainly a key to the pathology, possibly due to decreased osteogenic capacities and impaired local vascularization. Treatment of CPT is still challenging in pediatric orthopedics because of bone union difficulties, persistant angulation, joint stiffness and sometimes severe limb length discrepancy sequellae. Numerous treatments based on biological and/or mechanical concepts, surgical or not, have been reported with variable success rates. Vascularized fibular grafts and the Ilizarov technique have greatly transformed the prognosis of CPT. Despite these steps forward, repeated surgical procedures are often necessary to obtain bone union and the risk of amputation is never entirely eliminated. The effectiveness of new treatments (bone morphogenetic protein, bone marrow stromal cell grafts, pulsed electromagnetic fields, induced membrane technique...) still requires to be confirmed. Combining these new techniques with existing treatments may improve the final prognosis of CPT, which nevertheless remains poor.

Introduction

Of all the diseases in children, congenital pseudarthrosis of the tibia (CPT) is probably one of the most difficult to treat. Even today, failure to obtain bone union is frequent and the functional prognosis is mediocre because of residual deformities, joint stiffness and remaining length inequalities. The aim of surgical treatment is to achieve bone union of the pseudarthrosis while restoring alignment in the leg to prevent the risk of recurrent fracture and to preserve function and bone growth in the leg.

CPT is a rare disease in children, with an estimated frequency of 1/150,000 births. It can be defined as a disorder of the diaphysis which is revealed by either pseudarthrosis at birth or by a pathological fracture presenting in bone with modifications such as bowing, narrowing of the medullary canal or a cyst. Although its relationship with type 1

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neurofibromatosis (NF1) or von Recklinghausen's disease was confirmed in 1950 [1], the physiopathology of CPT has still not been clearly defined. This disease has many presentations, both clinical and radiographic.

Numerous mechanical and/or biological based approaches have been described in the literature to treat this disease with success rates that vary. The prognosis of CPT has changed considerably in the last few decades thanks to vascularized fibular transfers and the Ilizarov technique. Despite these advances, several operations are often necessary to obtain union of the pseudarthrosis, and the risk of amputation is never entirely eliminated.

Discovery and clinical diagnosis

Bowing may be discovered during a clinical examination at birth or in the first weeks in life which presents as bowing of the tibia convex anterolaterally or a discontinuity between the two bones of the tibial segment [2]. Severe neonatal forms or primary pseudarthrosis, in which signs are present at birth, can be distinguished from secondary pseudarthrosis, which is revealed by a pathological fracture when the child begins walking [3]. The clinical presentation varies considerably, from simple bowing to various extensive bone deformities causing more or less severe bowing in one or two bones of the leg, and which may result in a pathological fracture and pseudarthrosis (Fig. 1). Severity of shortening in the leg also varies.

Normally CPT is unilateral, located at the junction of the middle and distal thirds of the tibial segment with no predominance for sex or side. The fibula is also affected in more than half the cases. A complete clinical examination, in particular neurological and dermatological, associated with an investigation of the family history must be performed in the presence of limb anomalies in a newborn or an infant [2] to differentiate a diagnosis of isolated CPT from one of the bone anomalies associated with NF1. The latter is a multisystemic neurocutaneous disease with an autosomal dominant pattern and an estimated frequency of 1/4000 births. The diagnosis of NF1 is clinical, based on a series of criteria defined at the 1987 consensus conference [4]. These bone abnormalities correspond to primary dystrophic bone lesions, or are secondary to damage to the soft tissues, which affect bone growth. While the incidence of bowing and CPT is less than 4% in NF1 [5,6], half of the patients (40—80%) presenting with CPT are NF1 carriers [5,7].

Imaging

Standard X-rays show heterogeneous lesions from simple convex anterolateral bowing to true tibial discontinuity, with an image of resorption of the two ends of the fracture responsible for significant loss of bone substance.

Cystic forms are also found, in which bowing appears at between 6 weeks and 1 year. The cortices are continuous and condensed and thickened in the concave part of the curvature. The medullary canal is narrow and an image of a cyst can be seen at the apex of the curvature. The deformity gradually worsens until the cortex finally breaks, causing a transverse fracture [8]. In dysplasic forms, the bowing is visible at birth and sometimes pseudarthrosis may already be present. The tibia is narrow with an hourglass appearance and the medullary cavity is partially or completely obstructed. The fibula is frequently affected in these types. When pseudarthrosis has developed, the ends of the bone may be thin, atrophic or on the other hand, wide and hypertrophic. These radiological features define a certain number of criteria which are the basis of the different classifications of CPT.

The recent development of magnetic resonance imaging (MRI) will provide more detailed analysis of both bone and especially soft tissue lesions, in particular the periosteum

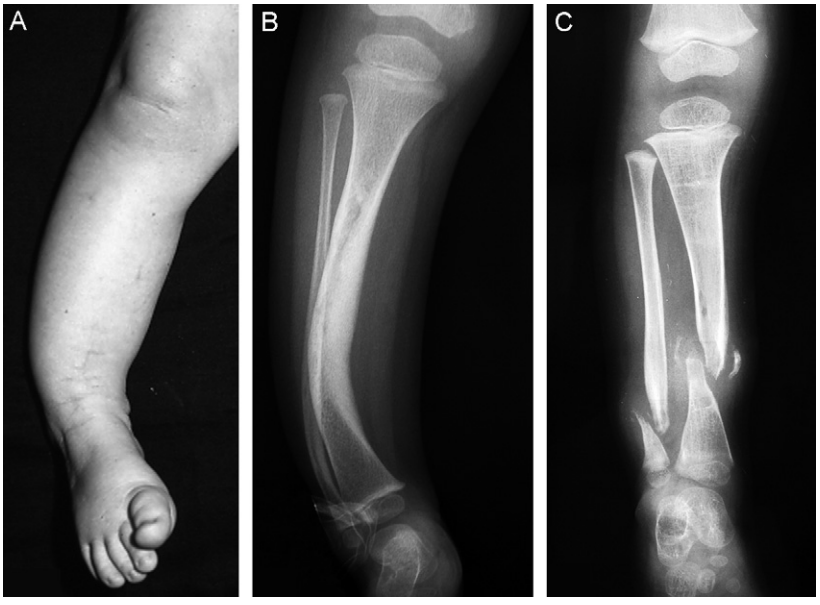

Figure 1 Clinical (A) and radiographic (B) heterogeneity of congenital pseudarthrosis of the tibia (CPT) with anterolateral bowing of the leg and atrophic pseudarthrosis of the lower third of the leg (C).