CLINICAL REPORT

Congenital pseudarthrosis of the tibia: An atypical proximal location

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Congenital pseudarthrosis of the tibia; Proximal location; Neurofibromatosis; Inter-tibiofibular graft

Summary
Proximal location of congenital pseudarthrosis of the tibia (CPT) is uncommon, and its management challenging, risking to end in amputation. We here report a case of proximal CPT managed in a limb-sparing perspective and followed up until the end of growth. A 17-year-old girl presented with type-1 neurofibromatosis and proximal CPT. Initial X-ray showed severe pseudarthrosis of the tibia with bone atrophy, 12-cm shortening and femorotibial and femoropatellar dislocation. Inter-tibiofibular graft and fibular tibialization were performed. At end of follow-up (age 33 years), fusion had been obtained. For orthoprosthetic and cosmetic reasons, a Boyd amputation of the tarsus was performed when the patient was 22 years of age. The functional result was very good, with 0—100° knee mobility. CPT, when proximal, completely disorganizes the knee joint, which is otherwise usually unaffected by this pathology. To achieve a good result, a limb-sparing treatment should combine correction of the tibial axis and of the dislocation of the knee, fibula osteosynthesis and bone graft.

Level of evidence: Level IV retrospective

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Introduction
Congenital pseudarthrosis of the tibia (CPT) is one of the main bone manifestations of Von Recklinghausen's neurofibromatosis (NF-1), present in 40 to 80% of cases [1,2]. According to Hefti's topographic classification [1], it is proximal in only 2% of cases, and there are in fact no studies that specifically concern such a location — which, however, has notable particularities, especially in terms of treatment.

The present report, of a case of proximal CPT followed up until the patient was 33 years of age, stresses the rarity of this location, highlights and discusses the treatment choices and pitfalls entailed, and presents results at very long-term follow-up.

Observation
CPT was diagnosed when the patient was a 9-month-old girl. Her right leg showed saber-like bending; initial X-ray
Congenital pseudarthrosis of the tibia

assessment (Fig. 1a) showed a defect in tibial diaphysis bone structure at the union of the proximal and mid thirds; coffee-colored patches were observed on the thorax, and a diagnosis of neurofibromatosis-related prepseudarthrotic curvature was made, retrospectively classified as Crawford type 2 [3]. Initial treatment (in a different center) was purely orthopedic, consisting in long-leg plaster casts, renewed until the age of 2 years, which failed to prevent evolution of confirmed pseudarthrosis. An unloading orthosis with ischial support, stabilizing the knee frontally while allowing flexion/extension, was prescribed until the patient was adolescent. Evolution was towards atrophic mobile pseudarthrosis (Fig. 1b, c). The patient reported no pain. Clinically, knee flexion was 115°, with a flexion contracture of 30° and 60° extension deficit and a 12 cm difference in limb length requiring a cumbersome, unsightly and relatively ineffective long-leg unloading orthosis with ischial support (Fig. 1d). There was a slight frontal laxity of the knee, hard to quantify due to the underlying mobile pseudarthrosis. Examination further found reducible and pain-free lateral dislocation of the patella. On X-ray, the CPT progressively migrated to the proximal third of the tibia, at 3.5 cm from the joint line, and was classified as Crawford grade 4 (Fig. 1b, c). It evolved into posterior subluxation of

Figure 1  a: first X-Ray at age 9 months: lateral concavity bending, involving the fibula; b: preoperative X-ray at age 17 years: extreme tibial atrophy with relative hypertrophy of fibula and femorotibial dislocation; c: preoperative X-ray of the knee with femorotibial dislocation; d: preoperative orthoprosthesis at age 17 years: neither esthetic nor functional, with one foot out of the prosthesis.

Figure 2  a: proximal inter-tibiofibular graft (immediate post-operative aspect); b: revision 9 months later for pseudarthrosis; c: revision for pathological fracture under the graft and pseudarthrosis evolution: distal inter-tibiofibular graft.
the tibia under the femur, with associated posterosuperior dislocation of the head of the fibula. Knee disarticulation was suggested when the patient was 16 years old, but was rejected by her family.

The patient was referred for treatment in our center at 17 years of age. She underwent two successive operations, the first being a proximal inter-tibiofibular iliac graft with an anterolateral approach (Fig. 2a), with long-leg cast immobilization. Nine months later, bone trophicity had improved thanks to tibiofibular synostosis, but the pseudarthrosis remained, requiring secondary intervention at the age of 18 years to tibialize the fibula and recenter the extensor system by transferring the tibial tuberosity. The pseudarthrosis zone consolidated without complication in 4 months (Fig. 2b). Two secondary complications, however, occurred after fusion had been obtained:

- a fatigue fracture of the distal quarter of the tibia, below the graft, occurred at 1 year and evolved into pseudarthrosis; it was managed by distal inter-tibiofibular graft at the age of 22 years, which secured consolidation (Fig. 2c);
- the ankle stiffened at a right-angle, leaving the forefoot hanging outside of the prosthesis.

This unsightly and dysfunctional situation was managed by Boyd tarsal amputation [4], with tibiocalcaneal arthrodesis, which enabled a brace to be fitted including the knee.

At end of follow-up, the patient was aged 33 years. She was satisfied with the result of the reconstruction, although judging it only tolerable from an esthetic point of view. Function was satisfactory (Fig. 3a, b). The knee was pain-free. Clinically, extension was complete, with 100° flexion. There was 15 cm shortening. There was a slight frontal laxity, with a good patellar axis but showing slight lateralization in the last 20° of active extension. On X-ray, fusion was good in both leg and ankle (Fig. 4a, b). The patient was socially integrated, studying law and playing sports (horse-riding and skiing).

**Discussion**

No literature reports focus on proximal CPT, which is considered exceptional. Hefti et al. [1] quote a frequency of 2%, with no indications as to treatment attitude. Lesion assessment at diagnosis can be difficult as, in nearly 29% of cases, topography varies over time, making the exact location unpredictable [1].

A proximal location is particular partly due to the impact on the knee and ankle. Without early surgery, the knee evolves towards femorotibial and femoropatellar dislocation with associated instability, and a pseudarthrosis area, which is itself mobile. As the leg needs to be suspended in a long-leg orthoprosthesis, the impact on the ankle can lead to stiffening and misalignment, disturbing overall limb function and hampering subsequent treatment.
It is difficult to decide exactly when surgery should be performed. Although osteolysis is more intense in early childhood, for some authors [5–13] surgery remains indicated at an early age, between 2 to 8 years. As some authors have reported consolidation rates approaching 85% after the age of 5 years, compared to 14% before [8–14], it would seem advisable not to operate too early, to avoid the pitfalls incurred by shifts in CPT and in intense osteolysis location, nor too late, so avoid the impairment of knee and ankle function mentioned in the present observation.

There are no treatment recommendations for proximal CPT in the literature. Various attitudes were reported to give varied results, as noted below, but which in all cases concerned medial or distal locations. The most complete data come from the European pediatric orthopaedic society multicenter study of congenital pseudarthrosis [8], with a cohort of 340 CPT patients from 13 countries. A wide and constantly developing range of treatment options is available to surgeons, and a review of the literature was able to shed light on respective results:

- Electrostimulation, mainly used in the early 1980s, seems to be the least satisfactory option in terms of consolidation, achieving a mean rate of 73% [5,7,13,15];
- two other, more widely used techniques — vascularized fibula transfer, and intramedullary nailing with graft— seem to give better results, with mean published consolidation rates of 92% and 88%, respectively [5,7,8,11,15–19];
- Ilizarov fixation stands out, with consolidation reaching 100% in some series [6,8,14,15,20,21];
- Limb amputation is resorted to in extreme cases [6], and especially in proximal CPT.

None of these techniques — and notably intramedullary nailing or vascularized fibula transfer, currently often used in diaphyseal involvement — were possible in the present case, due to proximity to the knee joint.

Evolution is often accompanied by complications, such as iterative fatigue fracture, at rates varying from 10.5 to 31% [20].

Boyd or Syme amputation [4] may be indicated in case of extreme shortening or when the foot prevents aesthetically and functionally effective bracing. The forefoot is then sectioned, with tibiocalcaneal arthrodesis, enabling compression of the pseudarthrosis and arthrodesis sites, with no significant problem of skin cover.

Finally, fibula status would seem to affect anatomic and functional prognosis [22]. 86.7% of patients in the EPOS study [11] presenting with ankle valgus showed fibular pseudarthrosis. The same authors claim that fibular involvement is a prognostic factor for good tibial consolidation [6]. In the present case, the fibula was hypertrophic and deformed, but without pseudarthrosis.

This case illustrates the difficulty of managing CPT in general, and proximal CPT in particular. Surgery should be early, long-lasting and conservative so far as possible, as amputation, if subsequently necessary, will require disarticulation of the knee. Consolidation should be ensured in every way possible, to minimize walking brace encumbrance, even though this will often require repeat intervention. Finally, enhanced comfort and esthetic aspect should be sought in every case in these child patients subjected to iterative surgery, with simple distal amputation when the often deformed foot begins to impair the function and esthetic aspect of the walking brace.

Conflict of interest

None.

References