**Congenital pseudarthrosis of the tibia**

**INTRODUCTION**

Congenital pseudarthrosis of the tibia (CPT) is a rare but challenging condition. The tibia shows area of segmental dysplasia resulting in anterolateral bowing of the bone. The osseous dysplasia leads to a tibial nonunion and, because of tibial bowing and reduced growth in the distal tibial epiphysis, shortening of the limb usually occurs. The disease becomes evident within a child's first year of life. The natural history of the disease is extremely unfavorable and once a fracture occurs, there is a little or no tendency for the lesion to heal spontaneously.

Treatment of CPT is still a challenge. The prime aims are to obtain a long-term bone union of tibia and fibula, to prevent limb-length discrepancy, to avoid mechanical axis deviation, soft tissues lesions, nearby joint stiffness and pathological fractures.

**SURGICAL OPTIONS**

CPT treatment is a difficult problem without clear solutions. Surgical treatment is not always efficient and even when bone healing seems to be obtained, new fractures can occur. The quest for durable bone healing could even deteriorate mobility and articular function. When articular function is compromised and when mechanical axis is deviated, amputation should be proposed to patients with CPT. Even if this solution allows fast functional recovery with the use of prosthesis, family often refuses it.

Depending on the type of treatment, age of the patient and severity of the CPT, the percentage of bone union varies between 31% and 100%. CPT healing rates were found to be higher when the pathological periosteum and fibromatous tissue were removed.

Moreover, esthetic and functional results are extremely variables. Treatment results should be evaluated once skeletal maturity is reached, growth is finished and deformity progression has ended.

Surgical options are vascularized fibular grafting alone or in association with external fixation or intramedullary rod and amputation for failed cases. The administration of BMP is still under investigation and preliminary results are encouraging. The efficacy of these methods remains to be proven.

*Age at Surgery*

The best moment to perform surgery is still debated. The results of a multicentre study conducted in 2000 by the European Paediatric Orthopaedic Society (EPOS) indicate a clear correlation between age at surgery and the final outcome, with better results being achieved in the older child. The EPOS report recommends surgery for CPT should preferably be postponed until the age of five and should not be performed on patients under the age of three years.

However, in a later report, Joseph et al. achieved consolidation in 92% of children under three years old and in 72% in children after three years old by external fixation. More recently, Pannier et al. and Gouron et al. reported union in patients as young as 14 months with all cases stabilized by a rod crossing the ankle.

*Vascularized fibular graft*

Vascularized fibular grafting, whether ipsilateral or preferentially contralateral, is a success overall as it permits initial tibia consolidation. However, many potential complications have been reported. Sometimes, secondary bone grafting is necessary to obtain bone union. Simonis et al. recommend using this method when limb length discrepancy is 5 cm-long and when pathological tissue length is greater than 3 cm. New-onset fractures are not rare, and they may be observed at the graft site extremity, or in the bone graft itself. Other complications include misalignment (anterior bowing and valgus deformity), as no remodeling occurs in these progressive angular deformities. Valgus deformity on the donor site has also been reported. In order to prevent this complication, distal tibiofibular metaphyseal synostosis (Langenskiold procedure) is recommended by some authors. However, this procedure may only delay, but may not prevent, the development of ankle valgus.

Patients from Montpellier, France were managed with the Charnley-Williams technique, adding to the insertion of an intramedullary rod into the tibia, a vascularized fibular graft according to the technique described by O’Brien. Initial consolidation occurred in every case and no fractures were recorded. The goal of this original technique is to obtain bone union, by mixing propitious biological environment with the vascularized bone graft, and the intramedullary rod is responsible for stability. The intramedullary rod offers good tibial alignment and prevents refracture. Hypertrophy and vitality of the bone graft promote bone union.

This association seems to guarantee long-term consolidation and correct tibial alignment. It may also prevent re-fracture in severe CPT because it mixes mechanical strength of the rod and biological quality of the graft.

*External Fixation*

Many authors use circular external fixation as the treatment of choice of CPT. External fixation allows total resection of pathological tissue, ensures stability regardless of the amount of resected tissue, and allows extension of the member, correction of axial deformations and full support immediately after the intervention.

The EPOS report found an overall rate of consolidation of 75.5% and a main age at the time of surgery of 7.5 years. These data are close to those from Ohnishi et al., as consolidation was obtained in 86% of patients, with a mean age of 4.5 years at the time of surgery.

Cho et al., reported re-fracture-free rate of cumulative survival was 47% at five years and did not change thereafter. The risk of re-fracture was significantly higher with a younger age at surgery (less than four years), low cross-sectional area of the healed segment, recurrence of tumor, associated with persistent fibular pseudarthrosis, residual ankle valgus, removal of an intramedullary rod, and noncompliance with bracing.

*Intramedullary rod*

The use of intramedullary rod for the treatment of CPT was suggested by Charnley in 1956; Coleman and Umber et al. popularized this procedure in the following years. Telescopic intramedullary rod with or without fibular fixation and classical rod with fibular fixation are variants of the original procedure that are associated with good results, as recent studies report an union of 80% or more.

Intramedullary rod of sufficient length must be selected to ensure that the tip of the rod reaches the proximal tibial metaphysis. Transarticular placement of rod across ankle provides more stable immobilization of small distal fragment. With use of non-elongated intramedullary rod, the rod will have to be changed as the child grows. Growth of the tibia depends on the distal end of the rod being located proximal to the ankle joint, and ankle motion is regained in most cases.

Intramedullary rod maintains internal splintage, prevents re-fracture and prevents progression of angular deformity of diaphysis; therefore, it is inadvisable to remove the rod after union.

*Bone Morphogenetic Proteins and Bisphosphonates*

Dysfunction of osteoclasts and abnormal proliferation of spindle cells may be the pathogenesis of CPT with insufficient osteogenic ability. Fibrous hamartoma cells maintain some of the mesenchymal lineage cell phenotypes, but do not undergo osteoblastic differentiation in response to BMP. They are more osteoclastogenic than are tibial periosteal cells. Birke et al. recently combined the use of bone morphogenetic proteins with bisphosphonates (pamidronate or zoledronic acid) as an adjunct to surgical intervention in 8 cases. Balancing catabolic response of bisphosphonates with anabolic action of bone morphogenetic proteins could improve healing rates. Primary healing occurred in 6 out of 8 cases. Key factors to achieve union in CPT include adequate resection of the dysplastic tissue, stable fixation and the establishment of the best biologic environment for bone healing. Only a long-term follow-up and the study of others cases can confirm or invalidate the durability of reported results.

*Role of the fibula*

Pseudarthrosis or hypoplasia of the fibula is associated with CPT in two thirds of the cases. Yet the need for fibular surgery remains controversial. Based on findings from Johnston, resection of fibular pseudarthrosis is necessary in order to achieve optimal limb alignment and union. If the fibula is intact, it is necessary to perform a fibular osteotomy. Dobbs et al. reported a higher rate of union in patients in whom fibular pseudarthrosis was resected as compared to those in whom fibular pseudarthrosis was not resected. Choi et al., classified fibula in two types: Type A (mild-A1, moderate-A2) includes a fibula of normal integrity in the presence of established atrophic-type CPT, whereas type B (mild-B1, moderate-B2, severe –B3) has atrophic-type CPT with concomitant fibular pseudarthrosis. They recommended ankle stabilization with end-to-end osteosynthesis of the fibula for mild (type B1), “4-in-1 osteosynthesis” (4 proximal and distal segments of the tibia and fibula are placed in 1 healing mass) for moderate (type B2), and distal tibiofibular fusion for severe (type B3) fibular pseudarthrosis in association with atrophic-type congenital pseudarthrosis of the tibia.

*Amputation*

For resistant pseudarthrosis when other extensive surgical procedures have not achieved a functional extremity, either due to persistent nonunion or due to dysfunctional angular deformity, shortening, atrophy and stiffness, the amputation is entirely appropriate.

While waiting for surgery, tibial alignment should be maintained with an orthosis. This is important for kids, as it allows them to walk, run and socialize.

*Ultrasound*

Okada et al. reported the use of ultrasounds for CPT healing. The authors reported a case of CPT of the tibia (Boyd type IV) successfully treated with low-intensity pulsed ultrasound stimulation (LIPUS) was administered for 20 min/day. The treatment was continued for 1 year until solid fusion on radiographs and subsequent full-weight-bearing. The underlying mechanisms of action of LIPUS remain unclear. However, in experimental studies conducted in rats, LIPUS application facilitates union and increase mechanical strength of bone.

*Complications*

Every surgical technique, including vascularized fibular grafting, external fixation and intramedullary rod, has pitfalls. Valgus ankle deformity on the donor side, re-fracture in the graft itself, recurrent non-union at one end of the graft site and residual limb-length discrepancy are not unusual after vascularized fibular grafting. Circular external fixation procedure is long lasting, complex and associated with a high risk of infection. Main limitations of the use of intramedullary rod include the risk for ankle stiffness (following long-term transfixation of the tibiotalar and subtalar joints), immobilization (following intramedullary rod), re-fracture (after removal of the rod) and disturbance of the proximal tibial physis.

*Management of complications*

Complications and failures often occur in CPT treatment. Management of complications can be challenging. These patients need a careful follow-up and residual complications need to be treated as they occur to avoid mal-alignment. Even patients with bone union may have compromised function secondary to residual deformities.

***Re-fracture****.* Achieving anatomic alignment of the tibia and fibula minimize the risk of re-fracture, so it should be the goal of all surgical procedures. Intramedullary rod and external bracing would offer more effective protection against re-fractures. Despite apparently solid clinical and radiographic union, re-fracture can also occur. There is a high rate of secondary nonunion following corrective osteotomy of the proximal or distal tibia. The initial treatment can be orthopedic but nonunion is frequent leading to surgery associated intramedullary rod, bone grafting, casting or Ilizarov external fixator and electro-stimulation.

***Malalignment of the tibia****.* Diaphyseal malalignment of the tibia (procurvatum or valgus deformity) are progressive and do not remodel. Retention of a rod across the previous pseudarthrosis should have biomechanical benefits. The deformities of the proximal tibia can be corrected with osteotomy and external fixator with normal morphology of the tibia. The deformity correction with osteotomy is contraindicated through dysplastic tibia morphology, as it can lead to fresh pseudarthrosis site.

***Limb length discrepancy****.* Residual limb length discrepancy following successful union is a major problem. Growth abnormalities of the tibia, fibula and the ipsilateral femur abnormalities are also noted with congenital pseudarthrosis of the tibia which include inclination of the proximal tibial physis, posterior bowing of the proximal third of the tibial diaphysis, proximal migration of the lateral malleolus. Shortening of lower limb-the affected tibia is slightly shorter than the normal side from beginning. Progressive shortening of the leg occurs as long as the pseudarthrosis remains ununited and also associated with repeated unsuccessful operations. Contralateral epiphyseodesis of the femur and or tibia can be done for expected limb length discrepancy less than 5 cm at skeletal maturity. Proximal tibial lengthening can be performed in children with expected limb length discrepancy more than 5 cm at skeletal maturity. Proximal tibial lengthening by distraction osteogenesis can be performed at the metaphyseal, physeal or subphyseal level. A good-quality regenerate without any complication can be possible with single proximal metaphyseal lengthening in children with normal morphology of proximal tibia. As, repeat lengthening and proximal dysplasic tibia were identified as significant risk factors of poor bone healing, chondrodiatasis or subphyseal lengthening of tibia or femoral lengthening may be better indicated when there is obvious proximal tibial dysplasia or a previous lengthening history. Single proximal tibial and /or femoral lengthening and contralateral epiphyseodesis is good option for expected limb length discrepancy more than 8 cm at skeletal maturity. Femoral lengthening or contralateral distal femoral epiphysiodesis avoid tibial complications but with the problem of knee displacement.

***Ankle valgus****.*  Altered growth of distal tibial physis and associated fibular pseudarthrosis are the main causes of ankle valgus.Residual ankle valgus deformity compromises functional outcome. Progressive ankle valgus is a problematic postoperative donor-site morbidity of a vascularized fibular graft in children. To prevent this complication, tibiofibular metaphyseal synostosis (the Langenskiöld procedure) has been recommended. Correction by additional surgical procedures achieves good alignment of the lower extremity. In distal tibia corrective osteotomy, care should be taken to perform osteotomy in metaphyseal area in patient with normal morphology of distal tibia and to rigidly immobilized after surgery by a cast or an Ilizarov external fixator. If there is enough growth remaining, the easiest and safest method to correct ankle is to perform distal tibial medial hemi-epiphysiodesis with a malleolar screw.

***Ankle stiffness***. Ankle stiffness usually progressively regresses once intramedullary rod is removed from ankle. Ankle stiffness may be kept minimized with revised fixation with keeping the ankle and subtalar joint free once unequivocal, sound union of the pseudarthrosis could be achieved. Pain secondary to degenerative changes of the ankle can be treated with limitation of activity and shoe modification. Severe pain may require ankle arthrodesis.

**CONCLUSION**

Basic principle of excising pseudarthrosis with diseased periosteum, stable fixation and protected weight bearing till skeletal maturity are effective method. Amputation must be reserved for failure to achieve union even after two or more good attempts, worse deformity than that produced by prosthesis. BMP holds considerable promise for the future. Healing rates in the complex forms of CPT remain uncertain and warrant larger prospective multicenter trials.