


Case Report

Congenital Pseudoarthrosis Tibia in Child with Neurofibromatosis Type 1 Treated with The Combined Vascularized Fibular Graft and Elastic Stable Intramedullary Nail (ESIN)

Sulis Bayusentono¹, Trixie Brevi Putri¹ 

¹Department of Orthopaedic and Traumatology, Faculty of Medicine, Universitas Airlangga/ Dr. Soetomo General Hospital, Surabaya, Indonesia

Correspondence should be addressed to Sulis Bayusentono, Department of Orthopedic and Traumatology, Faculty of Medicine, Universitas Airlangga/ Dr. Soetomo General Hospital, Jl. Mayjen Prof. Dr. Moestopo 6-8, Surabaya 60286, Indonesia. e-mail: sbsentono@fk.unair.ac.id

ABSTRACT

Background: CPT is a disorder characterized by nonunion tibial fracture that occurs spontaneously or after trivial trauma. Therapy usually includes surgical management and aims to gain lifetime bony union, avoid leg length discrepancy, and to prevent mechanical axis deviation, surrounding soft tissue lesion, joint stiffness, and pathological fracture. CPT remains to be a challenging orthopedic issue due to the difficulty to gain and to maintain the union and functional limb. The combined VFG and ESIN is one of our preferred method in Surabaya.

Case: A 13-year-old boy complained about crooked right lower leg since 1-year old, and fractured at 11 years old. In January 2014, the patient underwent ORIF ESIN combined with free VFG. The patient got bony union 24 months after the ORIF ESIN and VFG surgery. RUST modified scoring system showed a score out of 3, which signified that there was a radiographically significant union at the site of tibial pseudoarthrosis.

Discussion: The combined VFG and ESIN is to maintain bony union and stability. Intramedullary fixation offers a good tibial alignment and prevents refracture. Hypertrophy and the vitality of VFG support the bony union. The level of success rate in both primary and secondary union is a benefit shown after VFG therapy.

Conclusion: Combination of VFG after complete resection of tibial pseudoarthrosis and ESIN in this case report showed a success in achieving and maintaining the bony union, thus this therapy became one of the preferences as a suitable standard therapy that we use in our hospital institution.

Keywords: Congenital pseudoarthrosis of the tibia; Vascularized fibular graft; ESIN; NF 1; Human and Medicine

INTRODUCTION

Congenital Pseudoarthrosis of the Tibia (CPT) is a disorder in which non-union fracture of the tibia that develops spontaneously or after trivial injury. This condition usually found in the first two years of life and most cases do not exist congenitally.¹ This pseudoarthrosis is a rare

condition and the incidence varies between 1:140.000 and 1:250.000.² It is found in 5,7% of patients with Neurofibromatosis type 1 (NF 1), while 40% of patients with CPT have NF 1.³ NF 1 is an autosomal dominant disorder with incidence 1:3500 live birth.⁴ Before the making

of exact diagnosis for this disease, it has to exclude other conditions that clinically seen as pathological fracture in pediatric such as infection, rickets, benign or malignant tumors, and osteogenesis imperfecta.⁵

CPT remains a challenging problem in orthopedics. It is owing to the difficulty in gaining and maintaining the union, resulting in getting functional limb. This case is different compared to another fracture in children. Several cases of CPT ended with dissatisfaction in the family due to the failure of surgical techniques with the shortening, the deformity, and non-functional limb. However, some cases ended in amputation.^{3,6} Refracture risk increase as the younger the patient get the surgery (younger than 4-year-old), the small longitudinal plane from the healing segment, dysplastic tissue recurrence, persistent fibula pseudoarthrosis, residual ankle valgus deformity, intramedullary rod removal, and non-compliance in wearing the brace.²

Therapy is primarily being done with surgery and it aims to gain long-lasting bony union, to prevent leg length discrepancy, to prevent mechanical axis deviation, soft tissue injury, joint stiffness, and pathological fracture. Several preferences of surgical therapy are presented such as intramedullary nail, external fixation, and also many osteoinductive substances so far. Nevertheless, none of the methods is considered being more superior in gaining bony union significantly.²

Several studies about the efficacy of various CPT therapy including the morbidity, the financial view, the duration, and the effort of the patient are the main point in determining the preferred method, accustomed to the available

resource and environment.⁷ Various novel techniques are being introduced including Vascularized Fibular Graft (VFG). This case report shows the evaluation of the combined VFG and ORIF ESIN as a viable standard therapy in managing CPT cases inpatient with NF 1 in our hospital institution.

CASE REPORT

Patient, 13 y.o. boy suffered from the crooked right lower leg and inability to use the leg due to instability. The family history presented that it was known that from the paternal line, there was one member of the family with the bilateral crooked lower leg with no sign of fracture. The clinical picture of the patient is shown in Figures 1 and 2. There was post-operative scar on the anterior proximal lower leg, anterior bowing deformity of the lower leg, leg length discrepancy of 4 cm (in which the affected leg was longer), Galleazzi sign was positive (Figure 3). Moreover, the patient had the café au lait spots all over the body (Figure 4).

At the age of 1, he sustained the crooked leg and had been advised to get surgery in our hospital institution but his family refused. At 10 y.o., the patient fell from 10 m height and complained of pain on the right leg. He got a long leg cast for 2 months. After the cast was being removed, he fell again and complained of pain and instability. He got additional examination in the orthopedic outpatient clinic and being diagnosed as Neurofibromatosis type 1. In January 2014, the patient underwent his first surgery ORIF ESIN combined with free VFG in our hospital



institution. At first, the surgery performed excision of pseudoarthrosis through the anteromedial longitudinal incision. Lesion of 17,3 cm in length was excised (Figure 4). Then the donor was taken from contralateral fibula (Figure 5) and continued by fixation of the donor fibula and recipient tibia using ESIN (Figure 6). At the final stage, anastomoses were performed between donor peroneal artery and recipient anterior tibial artery.



Figure 1. (A) Patient seen from a frontal view and (B) Lateral view, with *leg length discrepancy* of 4 cm.



Figure 2. Galleazzi sign (+), affected side is 4 cm longer



Figure 3. Café au lait spots all over the body

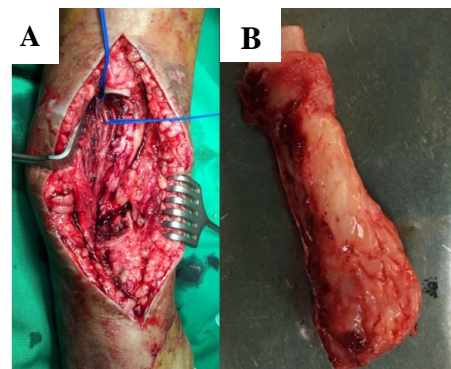


Figure 4. (a) Recipient tibia was cleared from (b) pseudoarthrosis tissue.

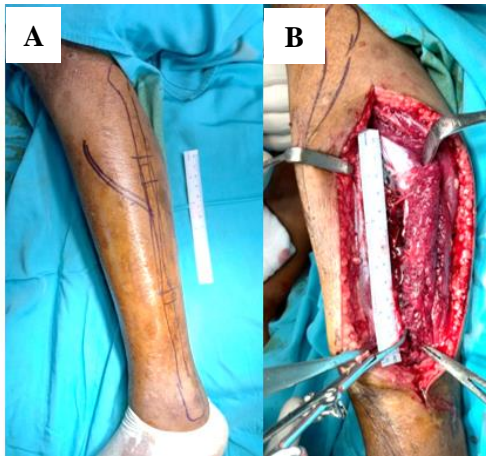


Figure 5. (A) Incision design to harvest the donor fibula from the contralateral leg and (B) measure the length of donor fibula to be harvested.



Figure 6. Fibula fixation donor to recipient tibia using ESIN

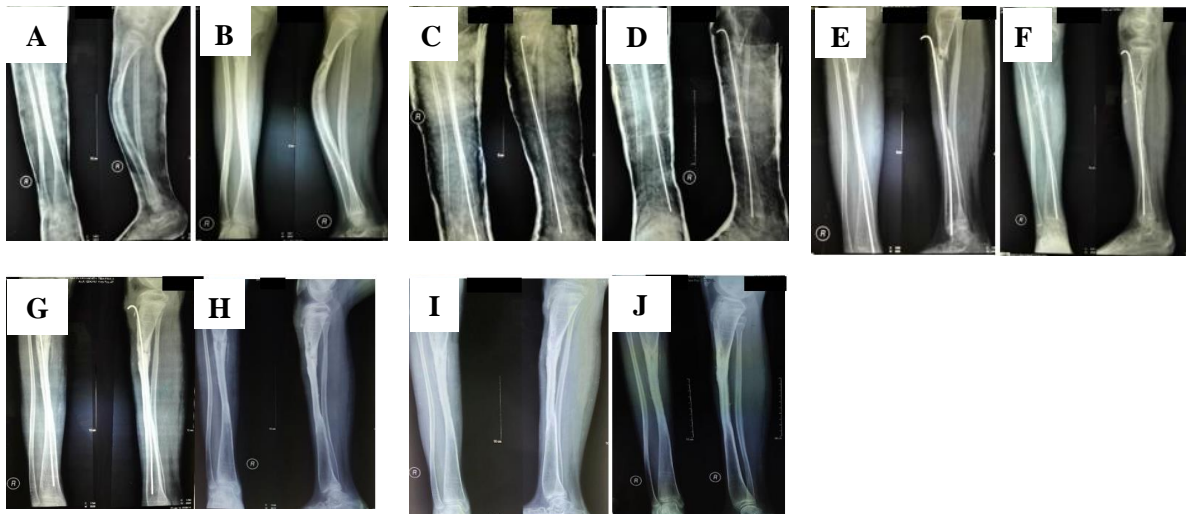


Figure 7. (A) AP and lateral view of the leg in May 2013 shows an incomplete fracture of the tibia on the proximal third with anterior bowing and angulation of 50° in cast application, (B) two months after casting, a radiolucent area could be seen (pseudoarthrosis lesion), (C) After VFG and ESIN in January 2014, (D) Eleven months after initial surgery, the ESIN revision was done to correct the alignment, (E) Elastic nail migrated away from proximal tibia, (F) After second elastic nail revision, (G) Twenty-five months after surgery, the alignment was good (mild angulation 8°) and *leg length discrepancy* 4 cm, (H) Radiological union existed, so the nail was removed, (I) Six months post nail removal, consolidation got more obvious, and (J) Five years after surgery, alignment was still good, showing remodeling process and there was no radiolucency/ pseudoarthrosis recurrency.

The patient had another 2 ESIN revision surgeries during 2014 – 2015 to correct the alignment and implant position in the bone medulla. The bony union was gained and the nail being removed in February 2016 (25 months after VFG and ESIN being performed). The patient wore the brace for 2,5 years. The sequential x-ray images were shown in figure 7. Five years after the surgery, the patient was admitted to the orthopedic outpatient clinic with the condition of 3 cm leg length discrepancy. There was none of the complaints from the patient due to any pain or limping (Figure 8).



Figure 8. Clinical appearance 5 years after surgery

DISCUSSION

This case is one of a very rare case of CPT found in the orthopedic outpatient clinic. Based on the radiological type, this case is classified into Crawford type II where we could find cortical diameter constriction. In this case, the location of the pseudoarthrosis is proximal tibia which is only 2% of all CPT based on Hefti et al.⁷ This case fulfills diagnostic criteria of NF 1 due to NIH, the presence of café au lait macules and supported by the fact of the genetic anomaly in family history. The usage of

vascularized bone graft first introduced by Ostrup and Fredrickson in 1974. VFG was implemented clinically in 1975 by Taylor, Miller, and Ham. While its usage for CPT treatment was described by Judet *et. al.* on 1978 and a good result was reported by Chen, Yu, and Wang in 1979, Weiland and Daniel in 1980, Pho *et. al.* in 1985, while the long-term result has not been published.^{2,8-10}

In a study by Sakamoto *et. al.* for the application of VFG for CPT, the bone consolidation rate reached 94% and the average time to achieve consolidation is 6,6 months without any refracture. Considering this result, VFG become the primary choice of therapy in their institution.¹¹

The outcome in CPT treatment is classified using Johnston criteria as grade 1, where there is unequivocal union with full weight-bearing function and maintenance of alignment requiring no additional surgical treatment; the grade 2 categorized by the appearance of the equivocal union with useful function (the presence of longitudinal cortical deficiency or transversal residual) and/ or any deformity (usually $>15^{\circ}$ valgus, procurvatum, or recurvatum), with the limb protected by the brace, which additional surgery required or anticipated; and grade 3 classified by the presence of persistent nonunion or refracture, requiring full-time external support for pain and/ or instability.³ This case report showed the complete union of CPT with ESIN and VFG, which is assumed to be the grade 1 classification. There is mild alignment radiographically (8° valgus and leg length discrepancy of 3 cm) where none of the



complaints (pain or limping) for the patient in this case.

In addition, the patient is free from the brace usage. The combined VFG and intramedullary fixation using ESIN is to gain bony union and stability. Intramedullary fixation offers good tibial alignment and prevents refracture. Hypertrophy and vitality of the vascularized bone graft (VFG in this case) supports the bony union to occur.² This therapy has a combined mechanical feature (from the nail/ rod) and the biological feature (from the graft). The high success rate to gain union either primary and secondary is the main goal of this technique.² Multicenter studies showed the primary union rate from this technique exceeded 70% and the increasing success rate as this was performed in elder children.⁸

The limitation of this technique is due to financial issues, technical difficulty, relative weak protection from refracture, failure in correcting the leg length discrepancy, limb, and ankle deformity simultaneously.² Another obstacle is the need for second surgery involving another graft procedure at the recipient tibia fibular graft junction to reach union at pseudoarthrosis. Refracture risk could be found especially at the recipient bone graft junction or the shaft of the fibular graft before the hypertrophy phase.⁸ Angular deformity will not undergo remodeling and this could get more progressive after VFG. The relationship between angulation degree and multiple surgeries been performed before wasn't concluded.

CONCLUSION

The main goal in treating pseudoarthrosis of the tibia is to gain union and to maintain it, while also optimize the involved limb function. Later, to minimize the angular deformity (ankle valgus), leg length discrepancy, to correct proximal fibular migration, and to prevent refracture.¹² Combined VFG after complete resection of tibial pseudoarthrosis and ESIN finally could be considered as a suitable therapy for this patient. Any follow up for these patients should be done until skeletal maturity is reached to identify and rectification of the sequelae after the primary healing.² Regular check-up in the outpatient clinic and KAFO brace is also advised to prevent refracture to some point.

REFERENCES

1. Horn J, Steen H, Terjesen T. Epidemiology and treatment outcome of congenital pseudarthrosis of the tibia. *J Child Orthop.* 2013; 7(2): 157–66.
2. Shah H, Rousset M, Canavese F. Congenital pseudarthrosis of the tibia: Management and complications. *Indian J Orthop.* 2012; 46(6): 616–26.
3. Patil SN, Rao PS, Yalamanchili RK. Residual Nonunion in a Case of Two and-a-half Year Old Child Congenital Pseudarthrosis of Ipsilateral Tibia and Fibula Treated by Intramedullary Fixation with K-Wire and Allogenic Cancellous Strut Graft: A Case Report. *Int J Med Res & Health Sci.* 2013; 2(2): 284.
4. Zhu G, Zheng Y, Liu Y, Yan A, Hu Z, Yang Y, et al. Identification and characterization of NF1 and non-NF1 congenital pseudarthrosis of the tibia based on germline NF1 variants: Genetic and clinical analysis of 75 patients. *Orphanet J Rare Dis.* 2019;14(1):1–13.
5. Stammers K, Cope S. Congenital pseudoarthrosis in a child with neurofibromatosis. *BMJ Case Rep.* 2015; 2015:1–2.



6. Khan T, Joseph B. Controversies in the management of congenital pseudarthrosis of the tibia and fibula. *Bone Joint J.* 2013; 95 B(8): 1027–34.
7. Hefti F, Bollini G, Dungal P, Fixsen J, Grill F, Ippolito E, et al. Congenital pseudarthrosis of the tibia: history, etiology, classification, and epidemiologic data. *J Pediatr Orthop B.* 2000; 9(1): 11-5.
8. Pannier S. Congenital pseudarthrosis of the tibia. *Orthop Traumatol-Sur.* 2011; 97(7): 750–61.
9. Erni D, De Kerviler S, Hertel R, Slongo T. Vascularised fibula grafts for early tibia reconstruction in infants with congenital pseudarthrosis. *J Plast Reconstr Aesthet Surg.* 2010; 63(10): 1699–704.
10. Iamaguchi RB, Fucs PMMB, Da Costa AC, Chakkour I. Vascularised fibular graft for the treatment of congenital pseudarthrosis of the tibia: Long-term complications in the donor leg. *Int. Orthop.* 2011; 35(7):1065–70.
11. Sakamoto A, Yoshida T, Uchida Y, Kojima T, Kubota H, Iwamoto Y. Long-term follow-up on the use of vascularized fibular graft for the treatment of congenital pseudarthrosis of the tibia. *J Orthop Surg Res.* 2008 Mar 6;3:13.
12. Paley D. Congenital pseudarthrosis of the tibia: Biological and biomechanical considerations to achieve union and prevent refracture. *J Child Orthop.* 2019; 13(2): 120–33.

