Original Article

Management of Congenital Pseudoarthrosis using Ilizarov Device–Delsuth, Oghara Experience

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Abstract

Background: Congenital pseudoarthrosis of the tibia (CPT) is a rare and very difficult pediatric orthopedic condition to manage. It occurs spontaneously or from a trivial trauma. It results from a dysplastic periosteum that is more osteoclastic and less osteoblastic in nature. Successful treatment targets excision of the dysplastic periosteum and replacement with viable one and bone graft ± bone morphogenic proteins to aid union. Stabilization of the union site with intramedullary implants is advised to prevent refracture. **Aims:** To highlight the outcome of treatment of CPT with the Ilizarov procedure, bone graft, and use of bisphosphonate (sodium alendronate). **Patients and Methods:** This is a retrospective study of five patients treated for CPT using the Ilizarov procedure, bone graft, and use of bisphosphonate in the Delta State University Teaching Hospital, Oghara in Delta State, Nigeria. Inclusion criteria were patients that had a fracture of the tibia ± fibula spontaneously or from minor trauma. Exclusion criteria were patients that had pathological fractures or fractures from significant trauma. The outcome was graded into good, fair and poor. **Results:** Five patients were seen in this study. The male/female ratio was 2:3. The age was 6–18 years (mean = 11.5 years). The limb length discrepancy was 6–12 cm (mean = 7.2 cm) pre-operation and zero after completion of distraction. There was the loss of some regeneration in two patients at the end of consolidation. Two patients had hypertrophic CPT, while three patients had atrophic. Union was good in four and fair in one. Complications seen were ankle stiffness bending of the regenerate and loss of length of regenerate. **Conclusion:** CPT is a rare condition worldwide that is associated with non-union and complicated by post-operation refracture or non-union has been successfully treated with a procedure that entails use of Ilizarov technique, bone graft bisphosphonate and support with intramedullary nails.

Keywords: *Congenital pseudoarthrosis, dysplastic periosteum, fibrous non-union, Ilizarov device, tibia*

Introduction

Congenital pseudoarthrosis of the tibia (CPT) is a very rare deformity that occurs in 1 in 250,000 live births.[1] It results from a dysplastic periosteum in which there are hyperactive osteoclast cells and less active osteoblasts cells.[1,2] The result is the development of a dysplastic tibia that fractures either spontaneously or from a trivial trauma.[1,2] In worse scenarios, the fibula is also affected. Fibrous non-union then ensues. Congenital pseudoarthrosis has been found in patients that have neurofibromatosis and in excised fibrotic tissue from the non-union site, histology has shown the presence of neurofibromatosis nodule.[2] For ease of management and prognostication, it has been classified by Boyd.[3]

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Lee *et al*[4] in their study on the pathogenesis of fibrous hamartoma in CPT discovered that the hamartoma cells resulted from aberrant growth of NF-1 haploinsufficient periosteal cells that failed to differentiate in terminal osteoblast cells and arrested at a certain stage. They concluded that any treatment model of CPT must address this factor for it to succeed.

Cho *et al.*[5] in another study disclosed that both fibrous hamartoma cells in atrophic CPT and normal tibia periosteum have BMP-2 and BMP-4 gene expression, suggesting that they are from the same source. When the hamartoma cells in CPT are exposed to bone morphogenic proteins (BMP), they express a low level of alkaline phosphatase in keeping with low osteoblastic activity while normal tibia periosteum expresses an increased level of alkaline phosphatase indicating increased osteoblastic activity. They claimed that this response may be one of the causes of this disease. In the same study, they also observed that there was more expression of RANKL mRNA than OPG mRNA in CPT which was in keeping with the increased osteoclastic activity of osteoclast cells.

Seebach *et al.*[6] posited in their study that the number and proliferative capacity of human mesenchymal stem cells are positively modulated in multiple trauma patients but negatively in atrophic non-union. This may explain why CPT fails to heal or easily refractures.

Gang-Qiang *et al.*[7] in their study of the comparative analysis of the concentration of peripheral blood mesenchymal stem cells (PBMSC) and blood flow during distraction osteogenesis in multiple trauma, monotrauma, and non-union in Sprague–Dawley rats showed that both PBMSC and blood flow increased more in multiple trauma than in monotrauma and least in non-union.

CPT has been difficult to treat over the years.[1-3] Several methods used have failed because of recurrence[2]. This has been traced to the dysplastic periosteum and poor blood flow.[2] All efforts at achieving success were targeted at excising the diseased periosteum, grafting the area with viable periosteum obtained from a non-affected site, adding bone graft or bone morphogenic proteins (BMP) to the site, improving blood flow, and supporting the healed fracture site until bone maturity was attained.[2,8-11] Intramedullary rods for the tibia and Kirschner wires are used for the fibula. Drugs like bisphosphonate have been used to halt the activities of osteoclast cells. Plates and screws with only bone grafts after excising the dysplastic tissues have been used to treat CPA with reports of recurrences. Recently, some experts have achieved varying successes with distraction osteogenesis with the Ilizarov technique.[2,8-11] Amputation has been reserved for failed treatment and those with severe shortening of the leg and deformity of the ankle joint.

Paley *et al.*[11] in their study of the pathologic nature of the disease devised a treatment protocol that was successful in most of the patients they treated. The treatment protocol entailed excision of the dysplastic pseudoarthrosis joint and surrounding diseased periosteum, docking the fragments, and applying harvested bone graft and viable periosteum from the non-affected sites around the docked fragments. They did distraction osteogenesis to correct limb length discrepancy using the Ilizarov device and stabilized the tibia and fibula with intramedullary implants. In addition, the activities of osteoclast cells were halted by means of bisphosphonates (zoledronic acid infusion).[11]

In this study, we want to present our own experience using the treatment protocol similar to that suggested by Paley *et al.*[11] in the treatment of congenital pseudoarthrosis in the Delta State University Teaching Hospital, Oghara, Delta State, Nigeria.

Patients and Methods

This is a retrospective study of patients treated for congenital pseudoarthrosis of the tibia in the Delta State University Teaching Hospital, Oghara, Delta State, Nigeria, over a period of 5 years, from June 2017 to May 2022. The hospital is a tertiary health facility in the South–South geopolitical zone of the country and serves as a referral center for orthopedic cases.

Information for the study was obtained from patient case notes and includes biodata, type of CPT, limb affected, site of the limb affected, number of previous surgeries had, treatment received, outcome of treatment, and any complication that occurred. The inclusion criteria were patients that developed fractures after birth spontaneously or from trivial trauma. Exclusion criteria were patients that had fractures from significant trauma, pathological fractures resulting from infection or tumor condition. Ethical clearance was obtained from the institution's ethical committee. Data were analyzed by simple statistical analysis using SPSS-18 package. The outcome was graded into good, fair, and poor.

Procedure

The patient was placed in a supine position under spinal and regional block. Routine cleaning and draping were done. The fibrotic and dysplastic periosteal tissues were excised from the non-union site from both the tibia and fibula. The tapered ends of both the tibia and

fibula were also excised. K-wire was threaded into the tibia and fibula marrow cavity, respectively. While the fibular edges were compressed, that of the tibia had a gap of 2.5 cm. Ilizarov rings, connecting rods, and k-wires were assembled and applied to the affected limb. Corticotomy of the tibia was done between the 1st and 2nd rings. Corticotomy was also done on the fibula proximally to enable bone transport to take place unhindered. A latency period of 7 days was observed. Distraction was done between the 1st and 2nd rings while compression was done between the 3rd and 4th rings to close up the bone gap in the tibia. Distraction was done at the rate of 360° 12 hourly, while compression was done at 180° 12 hourly. For the other two patients, after excising the dysplastic tibia and periosteum and fibrous non-union tissues, docking was achieved and distraction was done proximally at the corticotomy site at the rate of 180° 12 hourly. Distraction continued until the limb length discrepancy was corrected.

The patient was placed on antibiotics, analgesics, muscle relaxants, vitamins C and D, calcium tablets, and bisphosphonate (sodium alendronate).

The patient was discharged after two weeks of admission after the mother had learnt how to turn the device and clean it. She was seen in the outpatient clinic every 2 weeks initially and later every 4 weeks until the shortening was corrected. X-ray was done monthly to monitor the progress. At the completion of distraction, the patient ambulated on partial weight bearing until regeneration became consolidated. At this point, the device was removed and the leg was put in a cast. Ambulation continued. After consolidation was satisfactory from the check X-ray, the cast was removed and full weight commenced. The outcome was graded into good, fair, and poor.

Table 1: Clinical presentation

Patient AU P	Age (Years)	Sex	Duration	LLD	CPT Type	NF Nodule & Café lait spots
1.	6	M	4 years	6 cm	Hypertrophic	Present
2.	10	F	8 years	12 cm	Atrophic	Present
3.	18	F	14 years	6 cm	Atrophic	Present
4.	13	F	8 years	6 cm	Hypertrophic	Present
5.	11	M	5 years	6 cm	Atrophic	Present

LLD=Limb length discrepancy. NF=Neurofibrotic. All were of poor social economic status. The sponsor of patient 2 was a Good Samaritan, others were by relatives. Patient 3 had two previous unsuccessful surgeries before presentation.

Table 2: Treatment received

Patient	Treatment given	TX Duration	Outcome	Complications
1.	Ilizarov procedure	6 months	Good	Nil
2.	Ilizarov procedure	12 months	Fair	Ankle stiffness, loss of regenerate.
3.	Ilizarov procedure	6 months	Good	Loss of regenerate
4.	Ilizarov procedure	6 months	Good	Nil
5.	Ilizarov procedure	6 months	Good	Nil

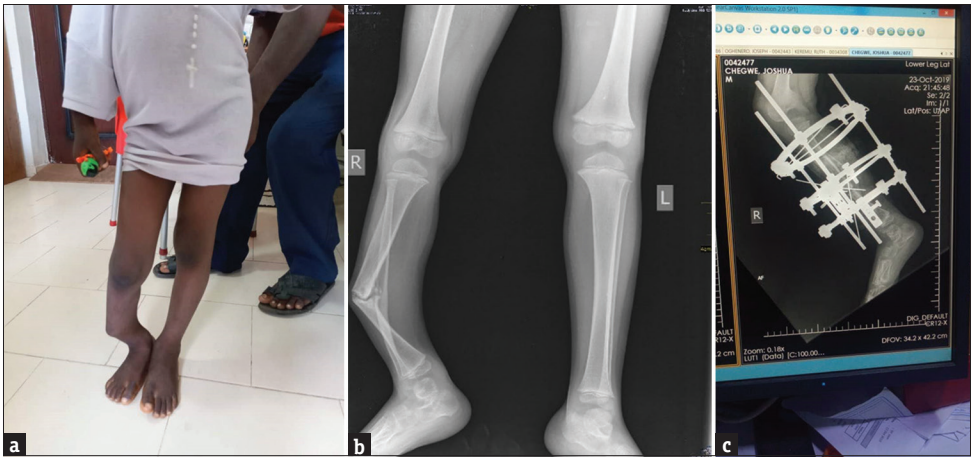

Figure 1: Clinical Photographs of Patient 1. (a) Before surgery. (b) Pre-op X-ray. (c) Post-op X-ray

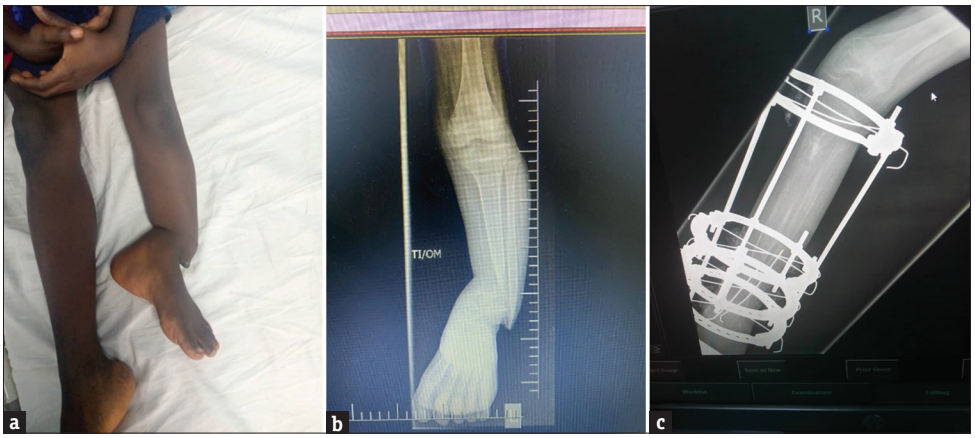

Figure 2: Clinical Photographs of Patient 2. (a) Before surgery. (b) Pre-op X-ray. (c) Post-op X-ray

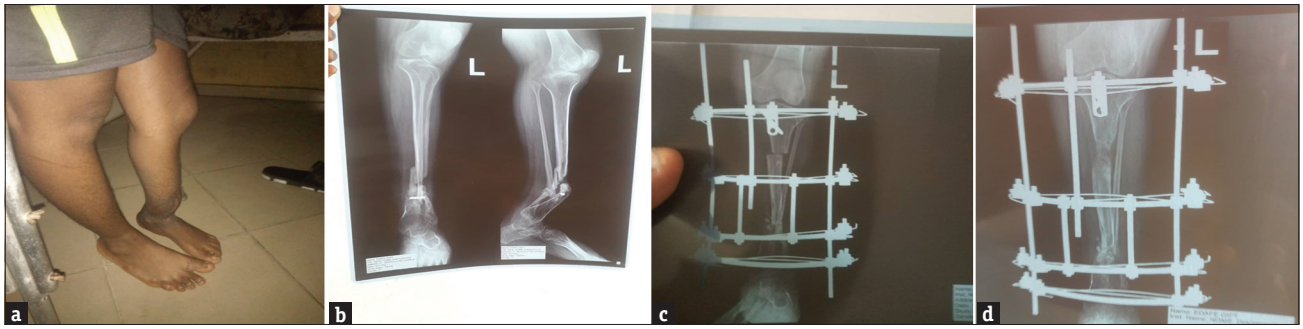

Figure 3: Clinical Photographs of Patient 3. (a) Pre-op photograph. (b) Pre-operation X-ray. (c) Post-operation X-ray. (d) Postoperative X-ray showing consolidation of regenerate and union at docking site of tibia

Results

Five patients were seen in this study. There was a male-to-female ratio of 2:3. The age range was 6–18 years with a mean of 11.6 years. The range of LLD was 6–12 cm with a mean of 7.2 cm. LLD was zero at the end of the distraction. All the patients had unilateral limb involvement and ipsilateral affectation of both the tibia and fibula occurred in three of the patients. All had café au lait spots. Two patients had hypertrophic non-union while three had atrophic non-union. The distal tibia was poorly developed in one of the cases seen. There was the loss of some regeneration in two patients at the end of consolidation. All the patients were of low social economic class. The treatment of four of them was sponsored by relations, while a Good Samaritan sponsored one. The outcome was good in four patients and fair in one patient.

Discussion

Congenital pseudoarthrosis is a very rare disease worldwide. This explains why the volume of patients seen in this study was low. The patients seen in this study were from poor socioeconomic class and their treatment was sponsored by either their relations or Good Samaritan. Patient 3 had two previous failed surgeries before presentation [Table 1].

All the patients are seen in this study presented with all the clinical features classical with CPT described in the literature. This made it easy to make a diagnosis. A histological diagnosis was still done from the excised fibrous non-union tissue from the pseudojoint to confirm diagnosis as expected routinely. We had both atrophic and hypertrophic varieties in this study [Table 1]. The atrophic type was more challenging to manage both surgically and during postoperative period. Seebach *et al.*[6] attested to this when they disclosed that atrophic CPT was associated with poor blood supply. Both atrophic CPT and the lengthy regeneration needed K-wire to stabilize the union site and prevent deformity at the regenerate level, respectively. This was in keeping with the observation by most authors.[2,8-11]

Three of the patients seen in this study had atrophic non-union and were in line with the findings of most authors. The two patients in this study managed by Ilizarov procedure did not have intramedullary support while the other three had. Umebese *et al.*[12] reported five cases of CPT. Four of the five cases had sclerotic CPT. They successfully treated them using intramedullary nailing without bone grafting after excising the pseudoarthrosis joint. In atrophic CPT, however, bone graft or bone morphogenic proteins is

needed to stimulate bone union after excision of the pseudojoint and surrounding dysplastic periosteum. The increased activities of osteoclast cells in atrophic CPT need to be halted in order to achieve bone union.[2,11] In this study, bone graft was used to aid healing at the docking sites. Paley *et al.*[11] used bone grafts while Eisenberg *et al.*[2] used bone graft and bone morphogenic proteins. Bisphosphonate (sodium alendronate) was used in this study to suppress the activities of osteoclast cells. Paley *et al.*[11] used bisphosphonate to manage CPT and achieved significant success in the cases they treated.

In this study, the Ilizarov device [Figures 1-3] was used to carry out distraction osteogenesis to treat this condition. Ilizarov technique enables the formation of new periosteum and neovascularization by distraction histogenesis, of which distraction osteogenesis is a part.[13] Neovascularization increases blood flow at the docked site.[13] This helps the docked tibia and fibula to unite and also supports the union achieved thereafter to prevent refracture.[13] Takazawa *et al.*[14] used free vascularized fibula graft (FVFG) inserted in the split tibia coaptation technique (STCT) to successfully treat two patients with CPT. This technique worked similarly to the Ilizarov technique to improve the blood supply at the excised pseudojoint to help the healing and maintenance of the union achieved. An external fixator was used in this procedure to support the tibia fragments during the union.[14] No intramedullary support was used in the two patients and they did not have any refracture after 7 and 4 years of follow-up, respectively.[14] This method, however, requires the expertise of a vascular surgeon and sacrifices the contralateral fibula in the process. The Ilizarov procedure does not need a vascular surgeon and does not involve the contralateral fibula.

The Ilizarov technique was able to correct the LLD in all of the patients at the end of distraction in this study, two of them, however, had a loss of some regenerate at the end of consolidation [Table 2]. An LLD of up to 12 cm was corrected, a fit not achieved by other methods of treatment (figures 2a,b & c). Umebese *et al.*[12] series had an LLD of 2.5 cm. Such patients would have had an amputation as the only viable option of treatment available previously. It addressed the valgus deformity of the ankle joint and also provided support for the ankle joint during ambulation after the LLD had been corrected while the union was going on and the regenerate was consolidating. This is similar to the support of orthosis [ankle foot orthosis (AFO) and knee ankle foot orthosis (KAFO)] used for conservative management of CPT in the early stages prefracture and the prosthesis used for patients that had Syme's or Boyd's amputation for failed conservative or operative treatment.[15,16] Syme's or Boyd's amputation is preferred to transtibial amputation before bone maturity is attained because of the complication of amputation stump bone overgrowth associated with transtibial amputation in growing children.[15,16] Plawecki *et al.*[10] successfully managed three patients with CPT with the Ilizarov device. They concluded that the Ilizarov device can correct both axial malalignment and LLD simultaneously.

Some authors have observed the significance of the poor osteopenic distal tibia fragment and deformed ankle joint in the poor treatment outcome of CPT. This has led to a lot of amputations and prosthetic fittings being offered as a last resort of treatment for such cases. This has been related to the poor blood supply in this region. Ilizarov, by neovascularization, improves blood supply to the site,[13] helps the fracture to heal, and so has reduced the number of amputations offered to such cases presently.

Paley *et al.*[11] and other authors pointed out the need to support the union site at the tibia and fibula with intramedullary devices. Eisenberg *et al.*[2] used a telescopic intramedullary device that lengthens as the child grows. Others have used solid Kirschner wires and Rush pins for the same purpose. In our study, Kirschner wires were used insome of the patients [Figure 2c].

The morbidities witnessed in this study were pains, muscle spasms, tearing of the skin, and pin tract infection. These were adequately taken care of with analgesics, wound dressing, and antibiotics.

The outcome of the treatment was satisfactory for all the patients treated and both patients and parents were very happy with it. They are, however, being followed up for recurrence and have been adequately counseled about the risk of it.

The drawback of this study is its low volume. Though this can be explained by the rarity of the condition, this is a preliminary report of our experience we intend to share. We wish to stimulate other surgeons to share their own experiences in the use of this treatment method of CPT.

In conclusion, CPT is a very rare condition worldwide which naturally ends up in non-union if untreated and even when treated, it is complicated by refracture or non-union. Ilizarov technique, bone grafting, use of bisphosphonate, and support by intramedullary nail have made the successful outcome of treatment of this condition possible.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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