DOI: https://doi.org/10.30795/jfootankle.2024.v18.1802

Case Report

Tibial hemimelia: a surgical approach for bone reconstruction and lengthening

Carlos Eduardo Cabral Fraga[1], Maria Eduarda Freitas Barbosa Arantes Vilela[1], Vinicius Ferreira Pires Bueno[2], Gustavo Teixeira Leão[1], Yesmin Naji Sola[3], Marcos Henrique Alves Resende[3]

1. Hospital das Clínicas da Universidade Federal de Goiás (HC-UFG), Goiâna, GO, Brazil.
2. Hospital de Urgência Otávio Lage de Siqueira (HUGOL), Goiânia, GO, Brazil.
3. Hospital de Urgência de Aparecida (HEAPA), Goiânia, GO, Brazil.

Abstract

Tibial hemimelia is a rare condition with a reported incidence of approximately one in one million live births. Although tibial hemimelia can vary its presentation, it often presents a shortened lower limb associated with deformities in the knee and ankle. The objective of this study is to present a case of tibial hemimelia and the surgical technique performed to correct the deformity and bone lengthening using a circular external fixator in several stages. A 15-year-old male patient was evaluated, presenting Jones type 3 tibial hemimelia (visible distal part, but no proximal) in the right lower limb associated with congenital clubfoot and ankle deformity. Studying and planning the patient's case was essential to classify and define the best treatment. These treatments can be flawed and often reach amputation. In the case described, several surgical approaches were performed with the objective of reconstructions and deformity correction, followed by limb lengthening, presenting excellent results.

Level of evidence IV; Therapeutic studies; Case Report.

Keywords: Hemimelia; Outcome measures; Tibia.

Introduction

Tibial hemimelia is a rare congenital pathology with an incidence of one in one million live births[1]. An important heritage correlation has been observed, with the condition linked to an autosomal recessive gene appearing unilaterally in 70% of cases[2]. Although tibial hemimelia can vary its presentation, it often presents a shortened lower limb associated with deformities in the knee and ankle. Initially, radiographs are requested for classification, indication of treatment, and prognosis. The first classification was proposed by Jones et al.[3] in 1978, which divides the deficiency into four main groups based on radiographs and bone morphology. Later, Weber[4] introduced a classification that considered the present cartilage, dividing it into seven types and 12 subtypes. Finally, in 2003, Paley[5] presented a new classification, modified in 2015, whose differentiation is directly related to treatment and prognosis[2].

Currently, the most used therapeutic approach remains amputation and prosthetics due to well-tolerated adaptation. However, recent studies and protocols have evolved to allow deformity correction and lengthening, preserving the limb, especially in mild presentations. In these cases, it is possible to use circular external fixators on the femur, tibia, and foot for limb stabilization and lengthening. In other cases, it is possible to apply Brown's procedure[6] to centralize the fibula between the femoral condyles and talus using gradual distraction for fibula centralization.

Study performed at the Hospital das Clínicas da Universidade Federal de Goiás (HC-UFG) Goiâna, GO, Brazil.

Correspondence: Maria Eduarda Freitas Barbosa Arantes Vilela. Rua 235 QD. 68 Lote Área, Nº 285, s/nº - Setor Leste Universitário, 74605-050, Goiânia, GO, Brasil. **Email:** dudavilela97@gmail.com. **Conflicts of interest**: none. **Source of funding:** none. **Date received:** June 22, 2024. **Date accepted:** August 20, 2024. **Online:** August 30, 2024.

How to cite this article: Fraga CEC, Vilela MEFBA, Bueno VFP, Leão GT, Sola YN, Resende MHA. Tibial hemimelia: a surgical approach for bone reconstruction and lengthening. J Foot Ankle. 2024;18(2):250-4.

The objective of this study is to present a rare case of tibial hemimelia, treatment with bone reconstruction using a circular external fixator in several stages, and to systematize knowledge and therapeutic possibilities.

Case presentation and surgical technique

A male patient presented with deformity in the right lower limb at birth and was diagnosed with a cavo-adducto-varus deformity at three days of life and started clinical treatment with serial cast immobilization for two months without satisfactory results. He was diagnosed with tibial hemimelia at six months; however, the patient was followed up in another service where surgical treatment was performed with tibia and distal fibula resection, talar dome associated with plantar fasciotomy, maintaining irregular follow-up.

At 16, he resumed follow-up with the persistence of congenital malformation, including cavo-adducto-varus, as well as deformity and joint instability in the knee and ankle. Additionally, there was a shortening of approximately 9 cm in the right lower limb, affecting both the thigh and leg (Figure 1).

Surgical treatment was planned in a single time, with tibiotalar synostosis and limb lengthening to correct dysmetria, and outpatient follow-up with lengthening presenting satisfactory bone regeneration, with programming for external fixator including the knee due to instability and continuity of treatment.

Surgical technique

The patient was operated on in dorsal decubitus, initially using the tourniquet. The first approach was anterolateral access to the ankle, with a slightly curved incision starting from 5 cm proximal to the lateral malleolus to 2 cm medial to the fifth metatarsal. Then, dissection was performed by layers until bone exposure of the tibia and fibula. A 4 cm resection of the distal portion of the tibia and fibula was performed, repositioning the right foot to a plantigrade shape and fixing it with three Kirschner wires. Once the tourniquet was removed, a new anteromedial incision in the proximal third of the tibia was performed for osteotomy in this same segment, following the Italian technique with multiple perforations with drill and irrigation with saline, completed with the use of an osteotome. After positioning a circular external fixator encompassing the hindfoot and tibial block to increase arthrodesis stability, these procedures were performed in July 2021 (Figure 2).

After the tenth postoperative day, the lengthening started at 0.75 mm/day (0.25 mm every eight hours) for one week, evolving to 1 mm/day (0.25 mm every six hours) until the end of the first month. In the second month, change the lengthening rhythm to 0.25 mm/day four times daily. Initially, the patient was resistant to the treatment using a circular fixator; however, he progressed slowly at the beginning of the third month, with partial weight-bearing using crutches. The radiographic follow-up showed distraction osteogenic with satisfactory bone regeneration, and the replacement of the fixator was scheduled to start the second stage of lengthening in December 2022.

In June 2023, the patient still resisted the total discharge of body weight on the limb, walking with compensation in footwear and crutches-no pain complaints, with strength deficit of the extensor mechanism. A new circular external assembly was then performed to finalize the lengthening, encompassing the knee in this second moment due to cruciate ligament agenesis. In addition, a new osteotomy of the proximal tibia was performed to allow distraction to continue (Figure 3).

After six months of outpatient follow-up, maintaining partial weight-bearing using crutches and lengthening with a distraction of 0.25 mm/day, it was reopened to remove the distal assembly in the femur and distal reassembly (Figure 4) in the hindfoot, allowing the patient to load and reinforce the formed bone regenerate (Figure 5). At this moment, the patient presented a stable right knee joint for daily activities,

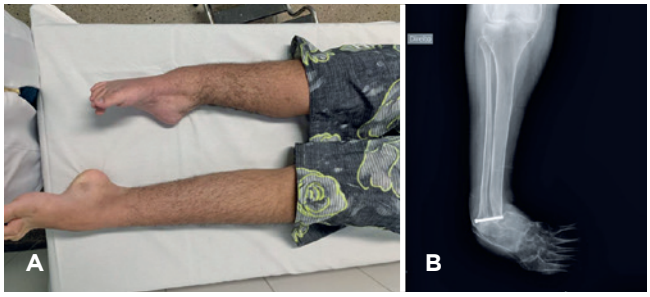

Figure 1. (A) Clinical aspect of deformity and shortening of the right lower limb (shortening of approximately 9 cm, with the right foot in cavo-adducto-varus) (B) anteroposterior radiography.

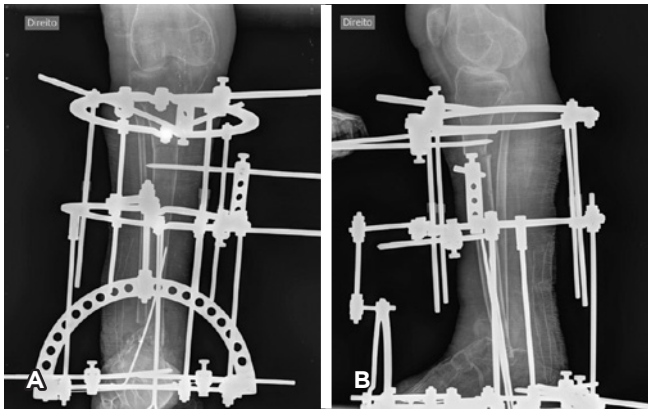

Figure 2. Anteroposterior (A) and profile (B) radiographs of the right lower limb after the first single-time approach of attempting talar tibial arthrodesis, fixed with Kirschner wire and assembly of a circular external fixator encompassing the tibial block and foot.

with a good range of motion, without pain complaints, with the plantigrade foot, allowing gait, however, with the presence of residual cavus.

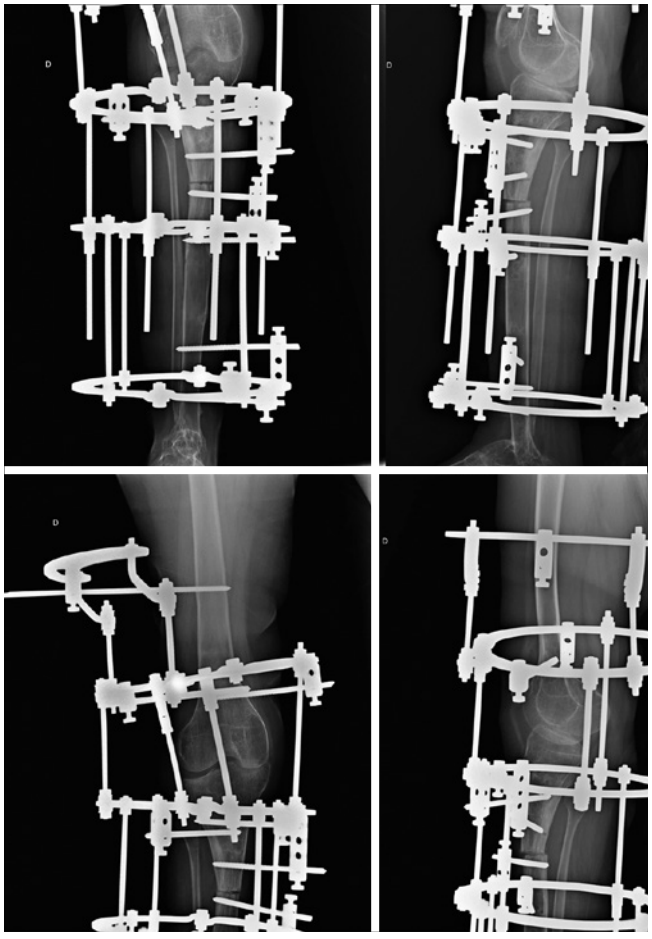

Figure 3. Anteroposterior and profile radiographs of the leg and knee, with the assembly of circular external fixator, including knee and ankle for bone lengthening, with osteotomy in the proximal tibia.

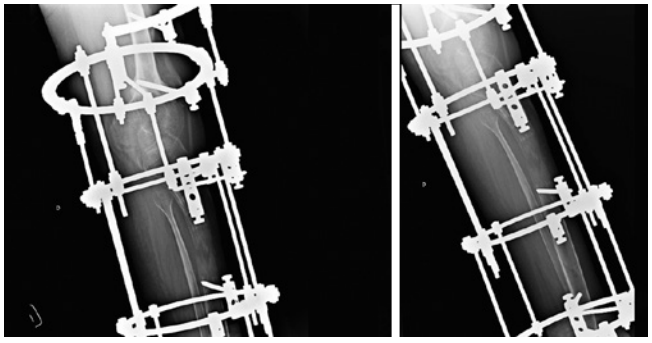

Figure 4. Internal and external oblique radiographs of the knee and right leg show bone regeneration in the formation process in the proximal third of the tibia after outpatient follow-up.

Discussion

The first case of tibial hemimelia was described in 1841; by 1941, there were already 79 documented cases[7]. Prenatal diagnosis can be made from the 16th week of gestation by ultrasound, and the genetic inheritance can be autosomal dominant or recessive.

There are different classifications for tibial hemimelia; Jones[3] is one of the most popular, using simple radiographs to differentiate into four main groups and 11 subtypes[1]. Type I has total distal tibia aplasia, subdivided into Ia, with distal hypoplasia of the femur, and Ib, with normal ossification of the epiphysis of the femur. Type II with ossified proximal tibia and distal tibial agenesis. Type III is the ossified distal tibia, and type IV is the shortened tibia with diastasis in the distal region between the tibia and fibula, as in the case above. The Paley classification[5], published in 2003 and modified in 2015[2], relates treatment to disease prognosis. The pathology is divided into five types and 11 subtypes, progressively evolving according to the involvement[2] (Figure 6).

Type 1: Hypoplastic tibia: valgus proximal tibial, relative growth of proximal fibula, tibial plateau present and normal**.**

Type 2: Proximal and distal tibial epiphysis present with dysplastic ankle

2A: Well-formed distal tibial physis; dysplastic tibial plateau; relative growth of proximal fibula.

2B: Delta tibia, proximal and distal growth plates connected through the epiphysis, ankle joint dysplasia; relative growth of proximal fibula.

2C: Delayed ossification (cartilaginous enlargement) of part, or all, of the tibia; dysplastic ankle joint; absence of tibial distal physis, relative growth of proximal fibula.

Type 3: Proximal tibia and knee joint present, medial malleolus present, distal tibial plateau absent, tibio-fibular diastasis.

3A: Lateral malleolus present, varus diaphyseal tibia, distal fibula with foot internally rotated around the tibia, talus can

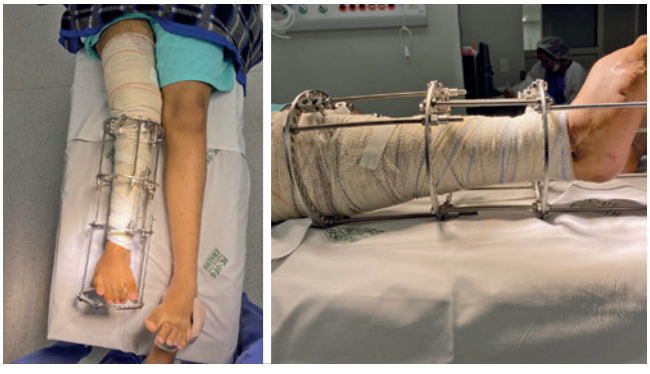

Figure 5. Clinical image after removal of the knee assembly, with a new distal assembly, encompassing the ankle to perform full weight-bearing on the right lower limb using crutches.

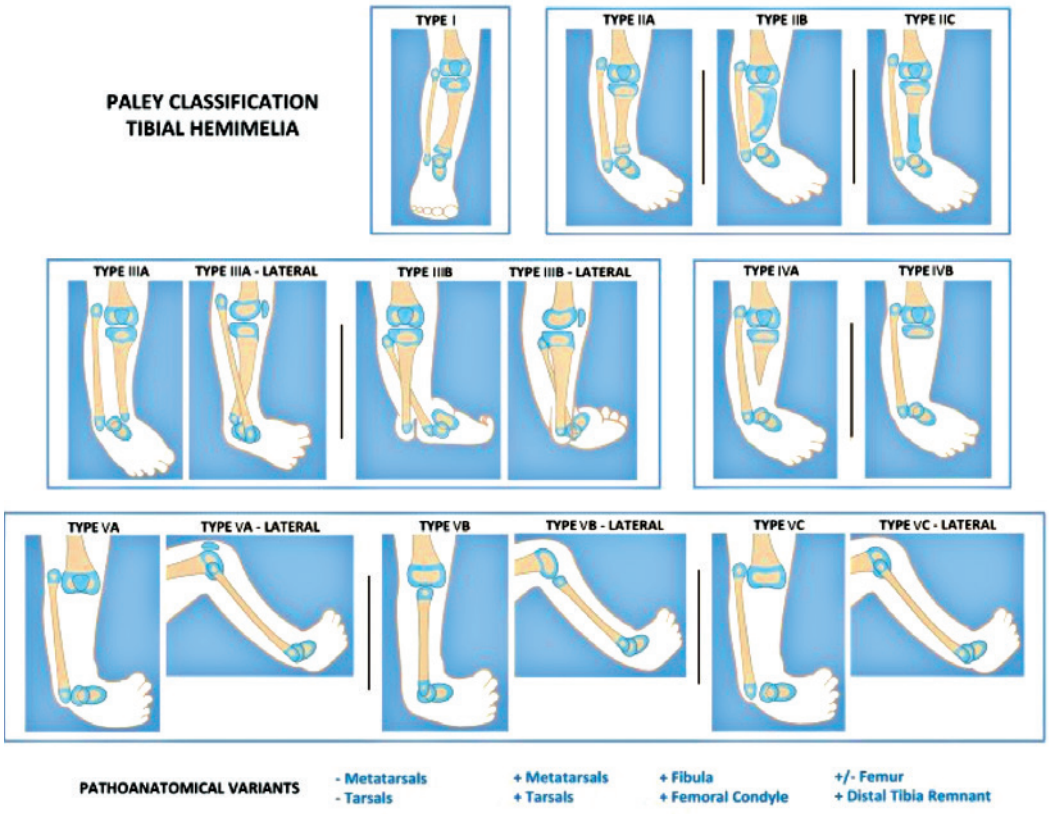

Figure 6. Paley Classification.
Source: Paley Foundation.

be positioned between the tibia and fibula due to absence of tibial plateau, relative growth of proximal fibula.

3B: Same as 3A with skin cleft separating the tibia and fibula, foot always follows the fibula.

Type 4: Distal tibial aplasia.

4A: Knee joint present, complete absence of tibia from diaphysis, pointed bone end often covered by skin pouch, relative growth of proximal fibula.

4B: Proximal tibial epiphysis present, knee joint present, relative growth of proximal fibula.

Type 5: Complete tibial aplasia.

5A: Complete absence of tibia, patella present; flexion contracture of knee, equinus-varus contracture of dislocated foot and ankle.

5B: Complete absence of tibia, no patella; flexion contracture of knee, auto-centered fibula, quadriceps present, knee capsule present.

5C: Complete absence of tibia, no patella; flexion contracture of knee, dislocated fibula, quadriceps present, no knee capsule.

In addition to the changes already described, they may present joint instability in the ankle and knee, malformations, and even muscular agenesis of the quadriceps and patella, cruciate ligaments, and dysplastic or absent collaterals.

The treatment of tibial hemimelia is controversial, with amputation being the gold standard, especially in Jones' subtypes Ia and Ib, arguing for faster adaptation, especially in younger people. However, treatment acceptance varies across cultures and bone reconstruction has become more common. Recent studies show that satisfaction and quality of life tend to be better with reconstruction, but complications such as contractures, instability, and infections occur.

The choice of reconstruction method depends on the classification, experience of the surgeon, and quality of soft and joint parts of the limb. In 1965, Brown[6] proposed a surgical technique for centralizing the fibula, with the purpose of transforming it into a functional tibia, combined with the Ilizarov technique for gradual correction of dysmetria and soft tissue distraction. In 2015, Paley[2] proposed an update to the classification, allowing new surgical approaches associated with existing techniques, including correction of foot deformity, femoral osteotomy, and patelloplasty when necessary, allowing improvement of soft tissues. However, the benefits and applicability of the treatment remain uncertain, especially regarding the persistence of contractures, the prolonged duration of treatment, and associated complications.

It is important to reinforce the decision between early amputation or correction. Studies state that patients who have been submitted to serial reconstruction procedures with prolonged use of the fixator experience improved self-esteem after correcting the deformity[7]. Subjective aspects such as decreased pain and a higher degree of satisfaction after length correction were also observed, presenting better psychological results. In the case presented, the previous approach and the patient's desire was the surgical approach for correction and stabilization of the ankle joint and bone reconstruction to correct deformities with a circular external fixator. At the end of the treatment, the patient presented an excellent result from the functional point of view, the right knee, despite the cruciate ligament agenesis, underwent good stability to demand daily activities, in addition to a wide range of motion, without evolving with residual knee flexion, an evolution described in some studies after the use of an external fixator encompassing this joint. A 7 cm length was obtained in the right tibia, with persistence of 2 cm of final discrepancy in the lower limbs. Regarding the foot, corrected until reaching the plantigrade position, allowing the discharge of total weight on the limb during the gait, using crutches, despite persisting with residual cavus, which will be later programmed and corrected. Finally, the patient preserved proprioception in the foot, which allowed greater stability during ambulation.

Final Considerations

Given the rarity and wide variety of presentations of tibial hemimelia, understanding its treatment becomes complex and challenging. Before choosing the best therapeutic approach, it is important to identify and classify the type of hemimelia, determining the prognosis and the best therapeutic options. There are several reconstructive techniques, but many end up failing and require amputation. In the case described, the patient had multiple approaches, using a circular external fixator for a long period, obtaining an excellent lengthening result, providing a plantigrade foot with satisfactory sensitivity and proprioception, in addition to allowing the functional quality of the limb.

Authors' contributions: Each author contributed individually and significantly to the development of this article: CECF *(https://orcid.org/0000-0002-3832-7397), and MEFBAV *(https://orcid.org/0000-0002-3003-2631), and VFPB *(https://orcid.org/0009-0003-4421-9549), and GTL *(https://orcid.org/0000-0003-3489-9192) Conceived and planned the activity that led to the study, wrote the article, participated in the review process; YNS *(https://orcid.org/0000-0003-0719-3223), and MHAR *(https://orcid.org/0009-0007-6711-0510) Data collection, bibliographic review; formatting of the article, bibliographic review; All authors read and approved the final manuscript.*ORCID (Open Researcher and Contributor ID).

References

1. Laufer A, Frommer A, Gosheger G, Roedl R, Broeking JN, Toporowski G, et al. Femoro-pedal distraction in staged reconstructive treatment of tibial aplasia**.** Bone Joint J. 2020; 102-B(9):1248-55.
2. Paley D. Tibial hemimelia: New classification and reconstructive options. J Child Orthop. 2016;10(6):529-55.
3. Jones D, Barnes J, Lloyd-Roberts GC. Congenital aplasia and dysplasia of the tibia with intact fibula. Classification and management. J Bone Joint Surg Br. 1978;60(1):31-9.
4. Weber M. New classification and score for tibial hemimelia. J Child Orthop. 2008;2(3):169-75.
5. Paley D, Herzenberg JE, Gillespie R. Limb deficiency. In: Staheli LT, editor. Pediatric orthopaedic secrets. 3rd. Philadelphia: Hanley & Belfus; 2003. p.406-16.
6. Brown FW. Construction of a knee joint in congenital total absence of the tibia (paraxial hemimelia tibia): a preliminary report. J Bone Joint Surg Am. 1965;47:695-704.
7. Chong DY, Paley D. Deformity Reconstruction Surgery for Tibial Hemimelia. Children (Basel) 2021;8(6):461.