

A rare case of tibial hemimelia, surgical technique and clinical results

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ABSTRACT

We report a nine-year-old boy with a type IIIa tibial hemimelia, according to the new Paley classification. We describe the x-ray findings, the surgical treatment technique, and the prognostic course of the patient. Descriptions of such cases are very infrequent in the literature and type of treatment is still object of debate.

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Introduction

Tibial hemimelia (TH) is a rare condition with a wide range of clinical presentations, ranging from a hypoplastic tibia to complete deficiency of tibia.¹ The incidence of TH is 1 in 1,000,000 live births.² Several authors tried to classify this pathology, they attempted to recommend proper surgical options for each types of TH.³ In 1861, Billroth firstly described the “tibial hemimelia”.⁴ This description was then modified by Dankmeijer in 1935.⁵ TH can occur for an autosomal dominant or recessive transmission.^{6,7} TH is associated with several syndromes such as Werner's syndrome,⁸ Langer–Giedion syndrome, tricho–rhino–phalangeal syndrome (TRPS II),^{9,10} tibial hemimelia diplopodia syndrome,¹¹ tibial hemimelia and split hand and foot syndrome¹² and tibial hemimelia micromelia trigonal brachycephaly syndrome.¹³ Other known potential cause is mother assumption of talidomide during pregnancy. Weber described a classification system for TH and recently Paley has performed a more accurate evaluation system.^{3,14} All these classifications are useful to recognize if tibial reduction defects or

cartilaginous anlage are present. Nevertheless, there is a lack evidence about the correct approaches according to the grade of TH. Therefore, types of treatment are usually demanding, even for expert surgeons. We present a case report, trying to discuss the anatomical findings associated with a rare type of TH.

Case report

A nine year old male presented with TH and clubfoot of the right leg (Fig. 1). According to Jones and Weber classifications, TH was a type II. More precisely there was a distal diastasis of the tibia and normal hip joint, normal femur, normal patella, dysplastic fibula, and normal muscle function (abbreviated *coI+/feI+/pal/tiII+/fiIII/pell*) with a score of 33 points according to Weber score system for TH. In addition fibula dysplasia was difficult to evaluate, in fact the third distal portion was thick as a tibia and articulated with talus (Fig. 2). On the other hand, the foot was internally rotated and equinus with a medial contracture. We considered this case as a

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Fig. 1. Pre-operative clinical picture of right affected leg.



Fig. 2. Pre-operative X-Ray evaluation of the affected leg. Talus joint with distal fibula and distal tibia is posteriorly placed.

Paley III a TH. Before surgery, we tried to mobilize medial soft tissues with serial casting without satisfactory results.

Due to the particular clinical setting of deformity, we decided to treat the patient using two surgical approaches to the ankle. Using the antero-medial approach (Fig. 3), we removed 4 cm of distal tibia (Fig. 4) and we lengthen the Achilles tendon with a Z-plasty technique. Soft tissues between tibia and fibula were gently dissected and moved from the syndesmosis. After this surgical step, we performed an antero-lateral approach to resect the distal portion of fibula and the dome of talus, to better achieve a functional ankle position. Before stabilization of ankle in proper position with K-wire, a tibiofibular synostosis was carried out using two screws (Figs. 5 and 6). Alignment of talus in both projections and closure of tibiofibular diastasis were checked using fluoroscopy. Post-operative X-rays demonstrated a good functional alignment (Fig. 7). We protected correction of deformity with cast bracing for 3 months. Weight bearing was not allowed for the first 45 days.

Discussion

Several classifications for TH have been proposed. Jones in 1978 firstly published his classification based on X-ray findings. Thirty years later, a new classification system was introduced by Weber taking into account X-ray and cartilaginous anlage.^{3,15}

According to Jones classification, our case corresponds to type IV. Jones divided TH in four types, ranging from the most to the last deficient. Type IV is shortened tibia with distal tibia-fibular diastasis. Considering the Weber classification, our case is type 2, “distal diastasis” of tibia and fibula”. For this type of TH, the incidence according to Weber is 5%.³ Weber also introduced a score system for TH, ranging from zero to 39; the higher the score, the less impairment grade suffered from the patient. Scoring patient's type of TH, we obtained 33. Paley et al recently has noted classification gaps between Jones and Weber system. They evaluated 113 THs according to Jones classification and they reported: 47 type Ia, 5 type Ib, 18 type II, 2 type III and 10 type IV. 31 cases (27.4%) were considered unclassifiable. On the other hand, following Weber classification: 18 type I, 11 type II, 3 type IIIa, 17 type IIIb, zero type IVa, 2 type IVb, 5 type Va, zero Vb, VIa, VIb, 4 type VIIa and 47 type VIIb. Using this system, only six cases (5.3%) were evaluated as unclassifiable. On this basis, Paley has introduced a new classification system developed to clarify treatment options and related prognosis.¹⁴ There are 5 types and 11 subtypes with modifiers to better represent TH associated deficiencies or duplications. Following Paley classification, our case is a type IIIa, reporting a deficiency of tibial plafond, medial and lateral malleolus still present, varus bowing tibia, relative fibular overgrowth and foot internally rotated with talus positioned between the tibia and fibula centered under the fibula. Paley found that if TH is classified using his system no unclassifiable cases are found.

Surgical treatments for TH are various. Depending on type of TH, surgical management could vary from different approaches such as amputation, leg reconstruction and correction of deformity, ankle arthrodesis, tendons lengthening or transpositions.^{6,16} In 1965, Brown published a new procedure for Jones type I providing fibular centralization.¹⁷ Observing his results, Brown introduced transposition of patellar tendon to the fibula and femoral shortening if needed to allow knee extension.¹⁸ Nevertheless, TH treatment is often challenging also for trained surgeons and frequently linked to poor results.¹⁹ Most surgeons think that through-knee amputation for Jones type I and below-knee amputation for other Jones types

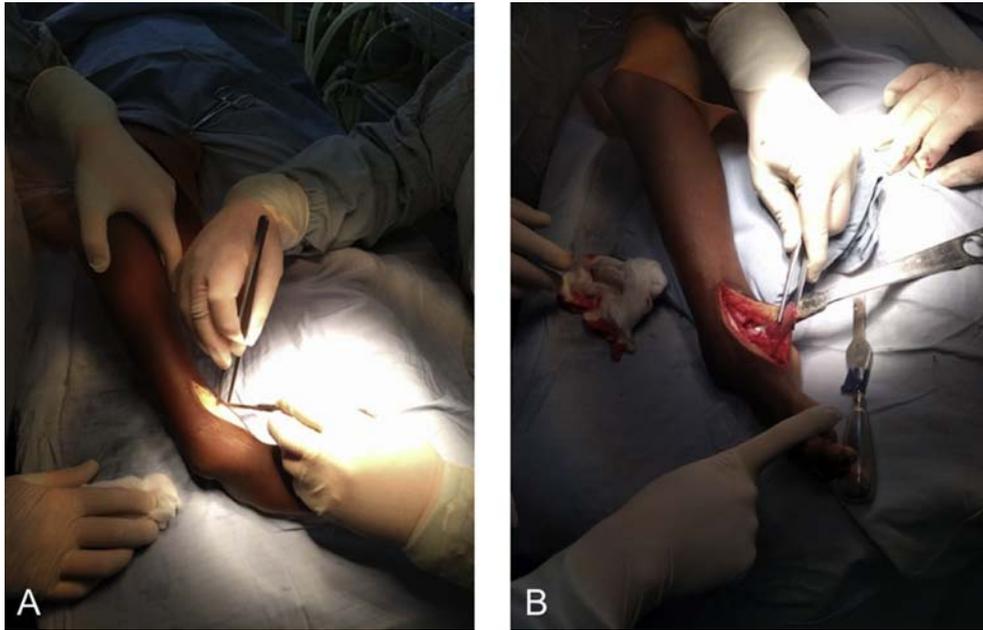


Fig. 3. Medial approach to the right ankle (A). This approach was used to isolate and resect the ex-ceeding distal tibial (B).

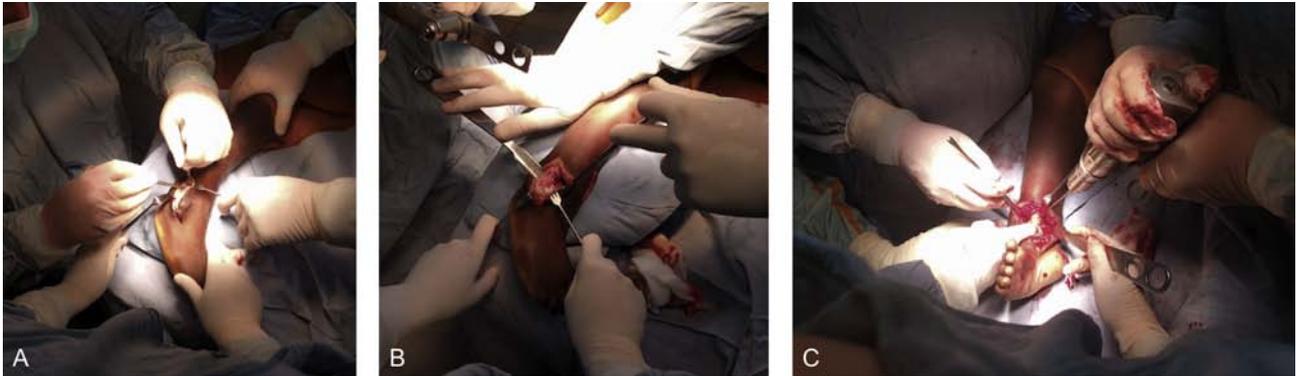


Fig. 4. The amount of tibial resection.

are the best treatments for TH. However, development of new surgical options such as hexapod frame offers different options as alternative to amputation.²⁰ Paley, in his recent classification,¹⁴ advocated different surgical treatments of several types of TH, explaining when an hexapod frames could be used. In Paley type IIIA, the foot could be gradually repositionated using these circular external fixators. He stated that if the talus is progressively distracted and positioned under distal tibial epiphysis, no fibular overgrowth resection is needed. For this circular hexapod frame, usually 1.5 mm wire is used to fix the bone to the rings. The proximal ring is attached to the tibia with one wire and two half pins, the distal ring is applied to the foot with one talar wire and three calcaneal wires. At the end of correction, when the foot is centralized, a second surgical step is usually needed to free the syndesmosis from the tibialis posterior tendon and to perform a syndesmotomic suture. Other wires are used to obtain partial epiphyseodesis of proximal and distal fibula and to avoid physiolyse

during distraction. In our case, since the patient was operated in Kenya, during a humanitarian medical mission, choice of treatment was conditioned by lack of resources. Since no hexapod external fixator frames were disposable, we aimed to treat the patient with further surgical steps, especially if a “biologic arthroplasty” or an ankle arthrodesis with tibial lengthening will be needed.

First of all, we aimed to reposition the foot under the distal tibia and fibula, reducing and compressing distal tibio-fibular diastasis with two trans-syndesmotomic screws and preparing the patient for a further ankle arthrodesis. A retrograde k wire was inserted to better stabilize the talus with the distal fibula. The other K-wire was used to ensure the foot in plantigrade position. We decided to maintain the plaster and the K-wires for 45 days. After this period, plaster was substituted with a weight bearing plaster and the patient was allowed to walk with crutches for other 45 days. After plaster removal, patient walked with an orthopedic shoe. Physiotherapy was performed to regain ankle range of motion.



Fig. 5. Lateral approach to the right ankle (A), this approach allowed us to resect the distal fibula and the talus dome (B). After this surgical step, we performed distal tibiofibular synostosis using two screw (C).



Fig. 6. Intra-operative picture of the correction of the foot. We stabilize ankle in proper position with two percutaneous K-wire.

Conclusion

Due to the rarity of the type of TH described, such case reports are very infrequent in the literature. Several classification systems



Fig. 7. Post-operative X-ray evaluation, latero-lateral (A) and antero-posterior view (B).

are commonly used. Paley classification best describes bone deficiency and patho-anatomy and help the surgeon to guide reconstructive options. Amputation option is likely used by the surgeons, especially for Paley types 2, 3 and 4 of TH or when modern surgical re-sources are not available. Reconstructive options for TH have improved in last decades. We advocate to attempt a reconstruction option first to better provide a functional lower leg with sensitivity and proprioception of the foot. Despite the lacking resources and the impossibility to use a hexapod system, we obtained a good clinical result, allowing preservation of the lower limb. For these patients, amputation option may be considered and overtaken in the future.

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