

Type-I Tibial Hemimelia

A Limb-Salvage and Lengthening Technique

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Background: Tibial hemimelia is a rare but disabling condition. Although reconstructive methods have been described, the recommended treatment typically has been amputation at various levels followed by the use of a suitable prosthesis. A new technique known as *femoro-fibulo-calcaneal arthrodesis* has been developed as a limb-salvage procedure for patients with type-I deficiency who refuse amputation or have no access to good prosthetic care.

Methods: Twelve children (18 extremities) with type-I tibial hemimelia were managed surgically. The ages of the patients at the time of surgery ranged from 2 to 14 years. The procedure includes 3 stages: loosening, lengthening, and stabilization. In all patients, the loosening stage involved release of soft-tissue contractures at both ends of the fibula. The lengthening stage involved either supervised lengthening at home with use with use of an external fixator (6 patients) or the use of traction in the hospital (6 patients). In all patients, the stabilization stage was subsequently performed by stabilizing the fibula to the femoral condyles proximally and the talus distally with use of crossed Kirschner wires.

Results: All 12 patients returned for follow-up for the first 5 years. All patients were evaluated by author. Two patients who had ipsilateral femoral focal deficiency were subsequently lost to follow-up. The remaining 10 patients were followed for a mean of 10 years (range, 5 to 32 years). All 10 patients were able to walk on their feet during follow-up. None of the patients in the present study had an amputation or needed a prosthesis. There were no major complications.

Conclusions: A new procedure, femoro-fibulo-calcaneal arthrodesis, has been proposed for the treatment of type-I tibial hemimelia. The suggested procedure is simple, biological, cost-effective, and dependable. It provides a long-term stable and functional extremity that enables the patient to walk with plantigrade feet with sensation and proprioception. This limb-saving procedure should be considered as an alternative for patients who refuse to lose the limb or for whom good prosthetic care is unavailable.

Level of Evidence: Therapeutic Level IV. See Instructions for Authors for a complete description of levels of evidence.

Tibial hemimelia is a rare congenital anomaly that may be associated with other congenital anomalies, deficiencies, and duplications¹⁻¹². The spectrum of pathological findings is much wider, and nearly 60% of patients with tibial hemimelia have other associated congenital anomalies such as lobster claw deformity, hand syndactyly, polydactyly, and/or foot ray duplication^{6-8,13,14}. Ipsilateral femoral bifurcation also has been reported^{2,15,16}. The fibula is always present⁸. The incidence of tibial hemimelia has been reported to be 1 per million live births^{5,17,18}. Parent-to-child transmission as well as families with multiple affected siblings have been reported^{9,13}. Nearly 30% of cases have been reported to be bilateral^{9,19,20}, and 72% have been reported to be on the right side^{11,19,21}.

In patients with Jones type-I tibial hemimelia, the quadriceps is deficient or absent and the knee and ankle are unstable with flexion contracture or dislocation^{1,3,8}. The patella is absent or dysplastic. The ankle is in equinovarus, with the foot supinated. The knee and ankle are unstable^{5,8,10,11,14,16,18,20,22-24}.

Many reconstructive procedures have been advocated^{1,3,19,24-26}. However, the recommended treatment has been amputation followed by long-term prosthetic replacement^{8,17,19,25,27,28}. A new technique of *femoro-fibulo-calcaneal arthrodesis* has been described as a cost-effective technique that can allow these patients to walk on their own feet with sensation and proprioception. The procedure may be a useful alternative for patients who prefer limb salvage to amputation.

Disclosure: No external funds were received in support of this study. The **Disclosure of Potential Conflicts of Interest** form is provided with the online version of the article (<http://links.lww.com/JBJSOA/A87>).

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TABLE I Data on the Patients

Case	Sex, Age(yr)	Side of Involvement	Associated Deformity	Quadriceps Power	Foot	Duration of Follow-up(yr)	Remarks
1	M, 12	Left	Polydactyly left foot	Poor	Severely supinated	8	Walking without support
2	F, 2	Bilateral	None	Poor	Severely supinated	32	Spontaneous femorofibular fusion; patient working as a teacher in a school
3	M, 2	Left	Bilateral syndactyly hand	Poor	Severely supinated	5	Walking without support
4	M, 3	Bilateral	None	Poor	Supinated	8	Walking with support
5	M, 14	Bilateral	None	Poor	Severely supinated	8	Walking without support
6	F, 2	Right	Unilateral syndactyly hand	Poor	Severe equinus	8	Walking without support
7	F, 4	Bilateral	None	Poor	Equinus	8	Walking without support
8	M, 2	Bilateral	None	Poor	Supinated, equinus	6	Walking without support
9	M, 2	Right	Toe duplication	Poor	Supinated	8	Walking without support
10	M, 5	Bilateral	None	Poor	Supinated, equinus	9	Walking without support
11	M, 4	Right	Congenital short femur	Poor	Supinated, equinus	Lost to follow-up	Lost to follow-up
12	M, 6	Left	Congenital short femur	Poor	Supinated, equinus	Lost to follow-up	Lost to follow-up

Classification

Various classification systems have been proposed to describe the types of tibial hemimelia^{3,8,27,28}. Jones et al. described a classification system with 4 types in 1978²⁸, and Kalamchi and Dawe suggested minor modifications to that system in 1985³. Weber described a new system with 7 types and 12 subtypes in 2008²⁴, and Paley described an extensive system with 5 types and 11 subtypes in 2016⁸. These systems have been used to guide prognosis, treatment strategies, and techniques for limb reconstruction.

We have followed the Jones classification system, in which types range from most deficient to least deficient. Type I is divided in 2 groups: type Ia (characterized by the total absence of a visible tibia) and type Ib (characterized by the presence of unossified proximal tibial epiphysis). Type II is characterized by an ossified proximal part of the tibia with distal tibial aplasia, type III is characterized by an ossified distal part of the tibia with proximal tibial aplasia, and type IV is characterized by a short tibia with distal tibiofibular diastasis.

Materials and Methods

The present study included 12 patients (18 extremities) who were observed at 3 teaching institutions between 1984 and 2016. Six patients had bilateral involvement, and 6 had unilateral involvement. The patients included 9 boys and 3 girls who ranged from 2 to 14 years of age. Nine extremities were involved on either side. Six patients had other associated anomalies: 1 each had bilateral syndactyly, unilateral syndactyly, polydactyly, and toe duplication, and

the remaining 2 had associated ipsilateral shortening of the femur and had been provided with a suitable prosthesis (Table I).

Preoperative assessment of all patients revealed a severe degree of flexion contracture of the knees. No patient agreed to amputation. One patient (2 extremities) had surgical correction of bilateral deformity. In the remaining 11 patients (16 extremities), lengthening was achieved along with the correction of the deformity. Postoperatively, the period of cast immobilization ranged from 16 to 24 weeks.

Surgical Technique

As the aim of the procedure is to achieve correction of the deformity and gain length, it has been considered essential to adopt 3 stages of reconstruction: (1) loosening, (2) lengthening, and (3) stabilization.

Loosening Stage

With the patient under general anesthesia and with use of a tourniquet, incisions are made over both the ends of the fibula. The severe soft-tissue contractures below the femoral condyles and above the talar dome are released. Both ends of the fibula are loosened without damaging the epiphyses. In cases of severe contracture at the knee, the origin of the gastrocnemius is also released and the Achilles tendon is tenotomized. In children, in whom the thickened fibula may be markedly curved, a double-oblique diaphyseal osteotomy may be performed. Two Steinmann pins are passed, 1 proximally through the lower end of the femur and the other distally through the talus.

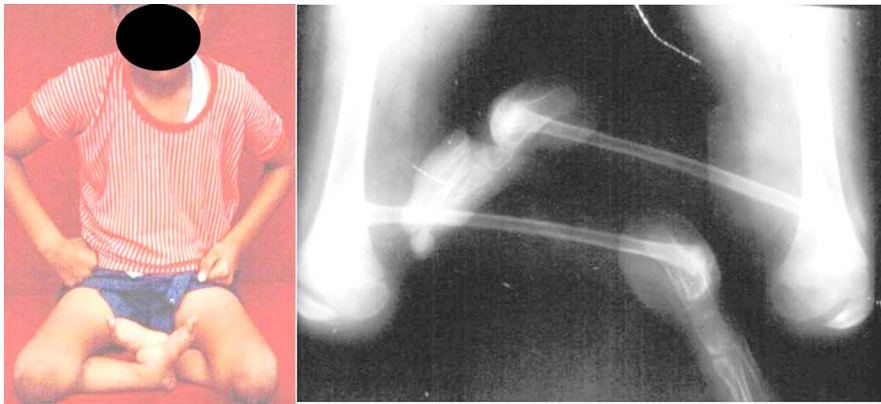


Fig. 1-A

Figs. 1-A, 1-B, and 1-C Case 1. **Fig. 1-A** Clinical photograph and radiograph made at the time of presentation.

Lengthening Stage

In children under the age of 8 years, a fixator is applied to achieve gradual lengthening. In older children, the limb is placed over a Bohler-Braun splint and traction is applied on either side (proximally on the head end of the bed and distally in a horizontal and balanced fashion). Half a kilogram of weight is added daily on either side under supervision. Once the desired length has been achieved, the fibula automatically becomes centralized, which provides shape and length to the leg. It is essential for the patient to be closely observed for any neurovascular complications during the period of lengthening.

Stabilization Stage

After the lengthening of the leg is achieved, the fibular ends are stabilized to the femoral condyles proximally and to the body of the talus distally with the help of crossed Kirschner wires. A foot-to-groin cast is applied. The deformity of the foot is corrected by means of posteromedial release. Neurovascular variations should be kept in mind when attempting to reconstruct the foot. In cases of bilateral involvement, the feet are corrected in the plantigrade position. However, in cases of unilateral involvement, 5° to 10° of equinus helps to compensate for the residual shortening of the limb. Weight-bearing is permitted in a snug-fitting cast.

Results

All 12 patients returned for follow-up for the first 5 years. All patients were evaluated by author. Two patients who had ipsilateral femoral focal deficiency were subsequently lost to follow-up. The remaining 10 patients were followed for a mean of 10 years (range, 5 to 32 years). All 10 patients were able to walk on their feet during follow-up. None of the patients in the present study had an amputation or needed a prosthesis. There were no major complications.

The extremities had spontaneous femoro-fibulo-calcaneal arthrodesis, and the patients were able to walk on their own feet. Only 1 patient needed cane support. We made a number of other observations. First, in all patients, the lower leg was markedly smaller than the thigh. However, during the follow-up period, the

fibulae in these patients demonstrated hypertrophy. Second, in the patients with bilateral involvement, the limb lengths were equal. Third, as the patients walked, the feet gradually gained good shape and size over time. Fourth, in patients with unilateral involvement, the technique was advantageous in gaining limb length and the remaining shortening of 1 to 3 cm was compensated with use of a shoe raise. Fifth, the hip movements were normal in all patients except the 2 cases of extremities with proximal femoral deficiency.

All 6 patients with bilateral knee arthrodesis learned to overcome the disability by activating hip movements.



Fig. 1-B

Radiograph made 9 months after the correction.



Fig. 1-C
Final radiographs of the lower extremities and clinical photograph of the patient, made 8 years after the correction of the deformity. The patient had bilateral toe deficiency.

These patients were functionally independent and were able to stand, walk, and perform activities of daily living.

In the present study, the technique resulted in a stable extremity with fusions at the knee and the ankle, no pain was reported, and patients could walk without the use of a prosthesis. No amputation was performed at any level, and there were no major complications. Given the severity of the initial anomaly, the results have been encouraging and the parents of these children have been satisfied. The last 2 cases in this series had ipsilateral femoral shortening and were managed with a suitable prosthesis.

Illustrative Case Report

A 12-year-old-boy presented for the treatment of a bilateral type-I deficiency, the diagnosis of which was confirmed radiographically (Fig. 1-A). He was managed with staged loosening, lengthening, and stabilization. The varus deformity of the foot was corrected in the plantigrade position. The leg was immobilized in a toe-to-groin plaster cast, and the patient was encouraged to walk (Fig. 1-B). The radiograph revealed femoro-fibulo-calcaneal fusion at 24 weeks (Fig. 1-C). The patient was able to walk without support.

Discussion

Tibial hemimelia is a challenging congenital anomaly that is associated with a wide spectrum of other congenital

defects and duplications^{1,8-11,16-21}. The condition is marked by shortening and bowing of the leg^{7,18,21,29,30}. To date, the reconstructive options have been limited, and the most accepted treatment has been amputation followed by a suitable prosthetic replacement^{1-5,7-9,17,21,22,27,31,32}.

It has been suggested that treatment should start as early as possible in order to achieve early and maximum alignment between the knee and the ankle^{5,17,30}. Various reconstructive methods have been described to avoid amputation^{1,12,19,24,27,30}. Brown, in 1965, described the cases of 3 patients who were managed with fibular centralization combined with arthrodesis of the knee and Syme amputation¹. Other authors have also recommended centralization of the fibula as a necessary step for reconstruction of the extremity^{1,8,17,25,26,30,31}. The technique proposed in the present study is simple and dependable. After loosening of both the ends of the fibula, centralization of the fibula automatically takes place during the process of lengthening. The centralization results in perfect alignment for fixation with crossed Kirschner wires. As both of the fibular epiphyses remain intact, further growth of the bone may be possible⁸. As suggested in earlier reports, we do not recommend fibular osteotomy³². However, for patients with a severely curved fibula, a corrective osteotomy may be considered³³.

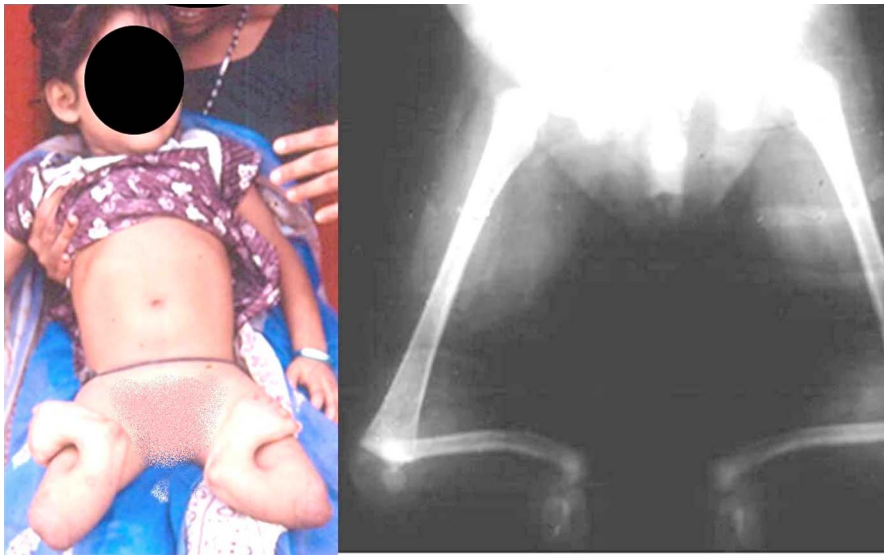


Fig. 2-A

Figs. 2-A, 2-B, and 2-C Case 2. **Fig. 2-A** Clinical photograph and radiograph made at the time of presentation.

Brown¹⁷, in 1971, and Jayakumar and Eilert³², in 1979, suggested reconstruction of the knee if a strong quadriceps is available. However, other authors have commonly observed that the quadriceps is either absent or poor in patients with type-I deficiency^{1,3,19,28}. Previous authors have also reported poor outcomes due to severe flexion contracture at the knee and the ankle and marked instability due to the absence of ligaments and poor quadriceps^{1,4,19,20}. Weber²⁷, in 2002, suggested patellar arthroplasty in cases in which the patella was present; in that procedure, the patella is fixed to the proximal part of the fibula to simulate the proximal part of the tibia. The soft-tissue release at the fibular ends as recommended not only helps with the centralization of the fibula but also permits relatively easy lengthening and alignment of the leg during the lengthening stage. The surgical detachment of the biceps femoris and both heads of the gastrocnemius plays a major role in the correction of the flexion deformity of the knee.

As both the knee and ankle are unstable in patients with type-I tibial hemimelia, it is not possible to achieve functionally mobile joints. Previous authors have described a number of different treatment options for such patients, including improved distraction and surgical techniques as well as staged reconstruction⁸; knee arthrodesis¹⁹; the creation of a stable, plantigrade, painless, fused ankle^{7,8,29}; and arthrodesis of both joints. The technique described in the present study simplifies treatment and can be carried out by most orthopaedic surgeons).

It is preferable to avoid knee arthrodesis, but the existing reconstructive procedures have not been very successful⁸. Although arthrodeses at the knee and ankle may seem a bit drastic, such treatment has been considered necessary in some cases. Patients who are so managed get used to the altered situation and remain active by using the hips to compensate for the distal arthrodesis in order to perform the activities of daily living. For example, one of the patients in the present study, who was 32 years old at the time of the latest follow-up, was married, had a child, was working as an

active teacher in a school, and was even able to go up the stairs, although slowly (Figs. 2-A, 2-B, and 2-C). Only 1 patient in the present study needed cane support.

In the present study, amputation or disarticulation was not acceptable to the parents and therefore the decision was made to attempt to salvage the affected extremities by means of



Fig. 2-B

Radiograph of the lower extremities, made 3 years after the surgical correction.



Fig. 2-C

Radiograph and clinical photograph made 32 years after treatment, showing perfect femoro-fibulo-calcaneal arthrodeses. Note the hypertrophy of the fibula. The patient was able to walk well and worked as a teacher in a school.

femoro-fibulo-calcaneal arthrodesis. The proposed treatment was readily acceptable to the parents. The advantage of this procedure is that it results in a stable and functional extremity that enables patients to walk on their own feet with sensation and proprioception. This procedure is a good option for patients who refuse amputation or have no access to good prosthetic fittings. None of the children in the present study had an amputation at any level, which has been a great source of satisfaction to the parents.

Conclusion

The results of the present study indicate that femoro-fibulo-calcaneal arthrodesis is an effective option for the treatment of type-I tibial hemimelia in patients who wish to salvage their extremity and wish to avoid long-term prosthetic fittings. This

one-time, cost-effective procedure results in a stable extremity and enables such patients to walk on their own feet with sensation and proprioception. ■

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