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Preliminary report on amputation versus reconstruction in treatment of tibial hemimelia

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Objective: Tibial hemimelia is a rare disorder characterized by the absence or hypoplasia of the tibia with associated rigidity. The aim of this study was to retrospectively evaluate the affectivity of reconstructive surgeries including centralization of the knee-ankle joints and lengthening with Ilizarov principles, as well as physical and functional results of amputation and reconstruction.

Methods: This is an IRB-approved retrospective review of all patients diagnosed with tibial hemimelia who required surgery at a single institution between 1998 and 2011. Charts were analyzed for clinical and radiographical findings. At final follow-up, patients underwent physical and radiographic examination. Patients and their parents were asked to complete the SF-10™ health survey (QualityMetric Inc., Lincoln, RI, USA).

Results: Twenty-one patients (12 male, 9 female) with 30 affected extremities were included. Mean age was 4.8±3.1 years at initial surgery. Knee level disarticulation was performed in 6 extremities of 4 patients. One patient with type III underwent transtibial amputation. Mean number of surgeries for each patient was 6.4±3.3, and mean duration of external fixator and casting was 17±6 months. Mean lengthening was 4.9±1.3 cm, and mean limb length discrepancy was 3.1±1.7 cm at 5.8±3.7 years at follow-up. SF-10™ scores were similar in disarticulated and reconstructed patients (p=0.63). All scores were significantly higher when disarticulation was performed in cases of knee instability (p<0.01).

Conclusion: When stability of the knee joint is present, treatment modality should be chosen according to the existence of the proximal tibia. Amputation should be preferred in cases of knee joint instability.

Keywords: Amputation; external fixation; knee centralization; reconstruction; SF-10™; tibial hemimelia.

Level of Evidence: Level III Therapeutic Study

Congenital deficiency of the tibia or tibial hemimelia is a very rare congenital deformity with a prevalence of 1 per million live births. It is a preaxial longitudinal deficiency with variable degrees of absence of the tibia. The fibula is usually intact, but there is aplasia or dysplasia of the tibia with marked shortening.[1–3] Flexion contracture of the knee, ankle joint instability, and dimple overlying the proximal tibial region are common. Rigid varus, supination deformity, and marked shortening of the first metatarsal are frequently associated with other medial ray defects (Figure 1).[4]

The Western literature suggests early ablative procedures for tibial hemimelia; however, this approach is not well accepted in many cultures, especially in Eastern

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Fig. 1. Clinical view of bilateral tibial hemimelia, flexion contracture of the knee, and ankle joint instability. [Color figure can be viewed in the online issue, which is available at www.aott.org.tr]

countries where orthopedic surgeons are pressured to perform reconstructive surgeries.[5] Distraction osteogenesis principles in both bone and soft tissue increase the expectancy of a functional extremity, even in difficult tibial hemimelia cases.

The most commonly used and accepted classification systems worldwide are those of Jones and Weber.[6,7] Although Webers' classification prioritizes soft tissue and anlage formation in callus, which is important in reconstruction surgery in tibial hemimelia, Jones' classification is widely preferred due to its simplicity.

Jones et al. classified tibial hemimelia into 4 types based on radiographic features.[6] In type IA, the tibia is completely absent, with a dysplasia of the distal femur. In type IB, the cartilaginous anlage of the proximal tibia is present, and distal femoral epiphysis is normal. In type II, the proximal part of the tibia is present, but the distal tibia is not visible. In type III, the distal part of the tibia is present. In type IV, the tibia is short, and there is distal tibiofibular diastasis with proximal displacement of the talus.[6,8]

The role of reconstructive surgery in severe tibial hemimelia (type I as described by Jones et al.) is controversial. Several studies recommend knee disarticulation rather than reconstruction for Jones' type IA in cases where there is total absence of the tibia and/or extensor mechanism of the knee joint.[4,9–12] Less controversy exists around incomplete or partial tibial hemimelia, with most authors in agreement that reconstructive surgery is appropriate.[6,9,10,13–15]

Although early amputation for congenital deficiency of the tibia is recommended, there are considerable advantages to retaining the foot in countries where people do not easily accept amputation.[4,8–11,16–18] Because of cultural and religious beliefs in Turkey, it is difficult to accurately survey views on amputation.

The purpose of the present study was to demonstrate the mid-term results of limb reconstruction in tibial hemimelia with external fixation techniques and compare the affectivity of treatment in different Jones' types. Limb length discrepancy (LLD), deformities, complications, activities of daily living (ADL), walking ability, and psychological status of patients were retrospectively evaluated in order to compare amputation versus reconstruction.

Patients and methods

This was an IRB-approved retrospective review of all patients diagnosed with tibial hemimelia who required surgery at a single institution between 1998 and 2011. Patients with a diagnosis of preaxial deficiencies which could not be classified by Jones' classification and patients with less than 2 years of postoperative follow-up were excluded.

Charts were analyzed for clinical and radiographical findings of associated anomalies, LLD, knee-ankle instability, functioning of quadriceps muscle, ray deficiency in the foot, total lengthening, and total number of procedures performed. Knee magnetic resonance imaging (MRI) was obtained in Jones' type I patients to assess the presence of cartilaginous proximal tibia.

Initial plain radiographs and MRIs (if present) were used to determine the type of tibial hemimelia according to Jones' classification by 2 observers retrospectively. Both observers were orthopedic surgeons with more than 2 years of experience in pediatric orthopedics and deformity reconstruction. Kappa coefficient of interobserver (0.92) was excellent for determining type of tibial hemimelia.

In type I and type II patients, treatment was first undertaken to address ankle joint instability and deformity. After posteromedial release, including achillotomy and posterior capsular release, the posterior facet of the calcaneus was centralized to the distal part of the fibula gradually with circular type external fixator (Figures 2a, b). Knee centralization was performed in combination with Brown procedure in type IA hemimelia, and knee-ankle joints (centralizations) were secured by a Steinman wire.[2] In type IB and type II, after the centralization of ankle joint, an osteotomy was made to the fibula

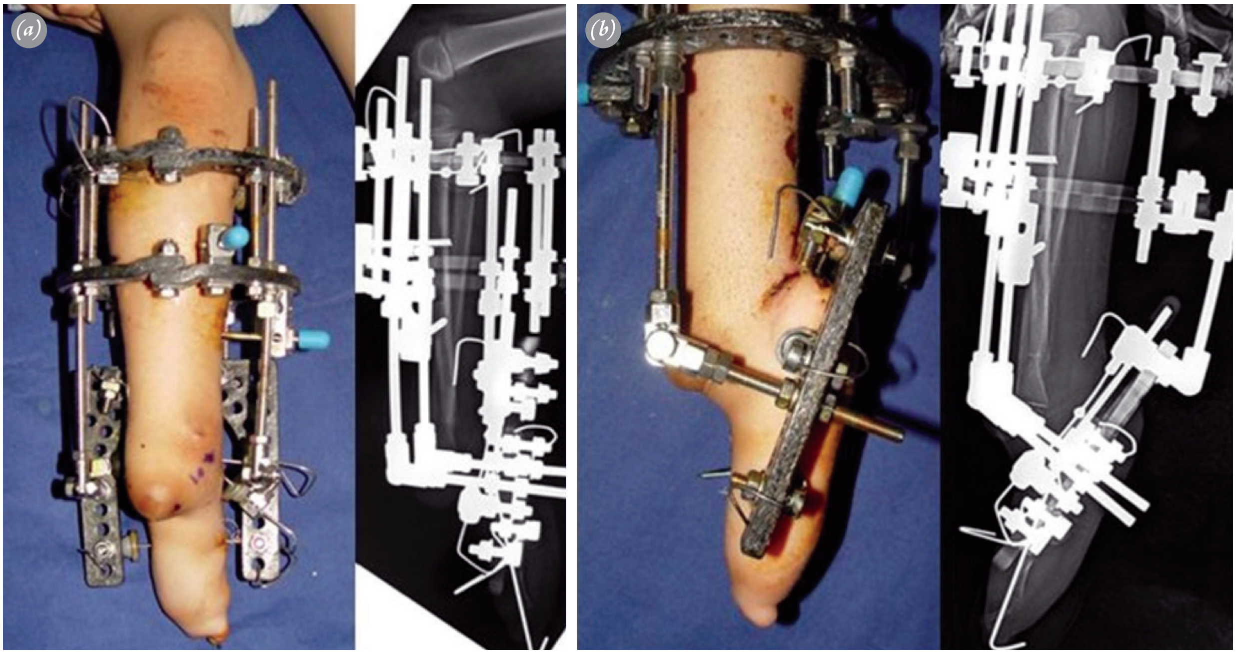

Fig. 2. **(a, b)** Clinical and radiologic view of gradual centralization of the foot to distal part of tibia in type II tibial hemimelia. [Color figure can be viewed in the online issue, which is available at www.aott.org.tr]

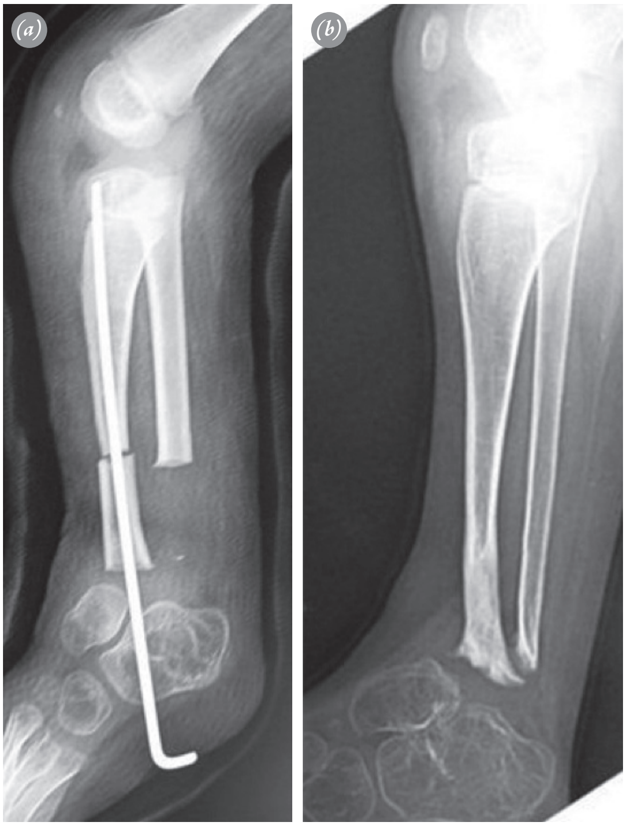

Fig. 3. **(a)** After centralization of the ankle joint, an osteotomy was made to the fibula at the most distal level of the tibia for the tibialization of the distal fibula. Ankle centralization and tibiofibular fusion were secured with a Steinnman pin through the calcaneus. **(b)** Consolidation of distal fibula to proximal tibia and regeneration of new ankle joint at 3-year follow-up.

at the most distal level of the tibia for tibialization of the distal fibula. Ankle centralization and tibiofibular fusion were secured by a Steinman wire (Figures 3a, b). In type IV, soft tissue release—including achillotomy and posterior capsulotomy—and distal fibula and tibia reorientation osteotomy were performed to close the diastasis at the mortis and achieve foot centralization. Osteotomies, arthrodesis, contracture releases, and lengthening procedures were applied in the remainder of the treatment process.

Deformities, LLD, complications, walking ability, joint instability, and stiffness were evaluated at final follow-up. Complications during reconstruction were classified according to Paley's classification system.[19] After physical and radiographic examination, patients and their parents were asked to complete the SF-10™ Health Survey for Children (Quality Metric Inc., Lincoln, RI, USA). The SF-10™ is a parent-completed survey that contains 10 questions adapted from the Child Health Questionnaire (CHQ). The SF-10™ provides coverage across a wide range of domains and is scored to produce physical and psychosocial health summary measures. This survey is intended for children between the ages of 5 and 18.[20,21]

Statistical analysis was carried out using Student's t-test for parametric data, Mann-Whitney U-test (Wilcoxon rank test) for nonparametric data, and chi-squared test for categorical data, as appropriate (SPSS v18.0, SPSS Inc., Chicago, Illinois, USA; Microsoft Ex-

Fig. 4. **(a)** Clinical and radiological view of lengthening and deformity correction for unilateral type II tibial hemimelia at different ages until the end of puberty. **(b)** 11-year follow-up. [Color figure can be viewed in the online issue, which is available at www.aott.org.tr]

cel 2013, Redmond, WA, USA). A p value of ≤0.05 was considered significant.

Results

Twenty-one patients (12 male, 9 female) with 30 affected extremities were included in this study. Mean age was 4.8±3.1 years (range: 1–12 years) at initial surgery. Jones' classifications[6] of patients are given in Table 1.

Different reconstruction procedures or different levels of amputation were applied according to subtypes of tibial hemimelia (Figure 4). All patients presented with ankle joint instability with or without knee instability, combined with deficiency of tibia. In patients with gradual knee instability (type IA), knee level disarticulation was presented to the family as a treatment option.

Knee level disarticulation was performed in 6 extremities of 4 patients. All disarticulated knees were Jones' type IA. One patient with type III underwent transtibial amputation. Sacrification surgeries were performed in early childhood, with a mean age of 53 months (range: 16–72 months). Four patients had as-

Table 1. Patient demographics and collected data.

Case	Gender	Side	Age at first operation	Jones' type	Associated anomalies	Joint instability	Initial LLD (cm)	Total lengthening (cm)	Total number of procedures	LLD at final follow–up (cm)	Age at final follow-up	Walking ability
1	M	R	3	IA	–	Knee, ankle	2	3	9	1	16	Walking with crutches
		L		IV	–	Ankle		4	5			
2	M	R	5	IB	–	Ankle	–	5	3	1	7	Walking with crutches
		L		IB	–	Ankle		5	4			
3	F	R	1	IA	–	Knee, ankle	–	4	5	–	6	Walking with brace
		L		IA	–	Knee, ankle		4	4			
4	M	L	2	IB	–	Ankle	3	6	9	2	6	Walking with brace
5	M	R	2	IB	–	Ankle	2	5	8	2.5	8	Walking with brace
6	F	R	3	II	Radial hemimelia	Ankle	3	7	12	4	16	Walking without assistance
7	F	R	8	II	–	Ankle	5	6	9	1	8	Walking with crutches
8	M	R	6	II	–	Ankle	4	5	8	5	16	Walking without assistance
9	F	R	5	II	–	Ankle	5	8	5	**	19	Walking with prosthesis and crutches
		L		IA	–	Knee, ankle		Disarticulation	7			
10	F	L	5	II	–	Ankle	4	6	9	3	11	Walking without assistance
11	F	R	1	II	–	Ankle	–	4	3	2	6	Walking with brace
		L		II	–	Ankle		6	3			
12	F	R	11	II	–	Ankle	6	6	12	4.5	15	Walking with brace
13	M	R	7	II	–	Ankle	–	7	7	2	9	Walking with brace and crutches
		L		II	–	Ankle		4	4			
14	M	L	2	II	Radial hemimelia, R vertical talus	Ankle	3	4	11	3	7	Walking with brace and crutches
15	F	R	8	IV	–	Ankle	6	5	9	5	11	Walking without assistance
16	M	R	1	IV	Radial hemimelia	Ankle	1	4	8	2	7	Walking without assistance
17	M	L	6	IV	Congenital short femur	Ankle	5	4	10	7	12	Walking without assistance
18	M	L	12	IV	–	Ankle	3	3	9	5	18	Walking with brace
19	F	R	2	IA	–	Knee, ankle	–	Disarticulation	1	–	4	Walking with prosthesis
		L		IA	–	Knee, ankle		Disarticulation				
20	M	R	4	III	–	Knee	1	Amputation*	1	**	5	Walking with prosthesis
		L		IA	–	Knee, ankle		Disarticulation				
21	M	R	6	IA	–	Knee, ankle	–	Disarticulation	1	–	9	Walking with prosthesis
		L		IA	–	Knee, ankle		Disarticulation	2			

*Transtibial amputation, **not available.

sociated anomalies such as radial hemimelia, congenital short femur, and vertical talus.

Mean number of surgeries for each patient was 6.4±3.3, and mean duration of external fixator and casting was 17±6 months. External fixator was used to correct ankle deformity, centralize the foot, and lengthen the effected extremity. Average lengthening was 4.9±1.3 cm by distraction osteogenesis.

Mean complications per patient was 5.5. There were 42 problems, including pin tract infection treated with dressing and oral antibiotics, and flexion contracture treated with physiotherapy. There were 16 obstacles. Regenerate fracture occurred in 9 patients: in 4 at the tip of the Steinmann pin, which was used to secure the centralization, and in 5 at the level of consolidation after lengthening. Debridement and pin removal was performed in 6 patients because of type III pin tract infection. Revision of external fixator was necessary in 8 patients during 3-dimensional foot transport and ankle centralization. Although revision of external fixator under anesthesia cannot be classified as an obstacle, any changes in the configuration of fixator increases the number of operations.

At time of publication, there have been 14 subsequent orthopedic events: 3 flexion contractures >30° at the knee joint, 2 equinus deformities, 3 knee dislocations, 2 knee subluxations, and 4 plastic deformations at the distraction level after removal of external fixators. Three of these plastic deformations required correction with re-osteotomy.

Mean follow up was 5.8±3.7 years, and average age at final follow-up was 10.3±4.5 years. Mean limb length discrepancy was 3.1±1.7 cm at final follow-up.

SF-10™ health survey scores were similar in disarticulated and reconstructed patients (p=0.63). Additionally, the scores were similar in unilateral and bilateral cases (p=0.12). Physical and psychosocial health summary measures were significantly higher when disarticulation was performed in cases of knee instability (p<0.01). SF-10™ scores were significantly lower in reconstructed type IA patients than in the remaining cohort (p<0.01).

At time of publication, all disarticulated and amputated patients continue to use prostheses. In the reconstruction group, 7 type I patients (100%), 4 type II patients (36%), and 2 type IV patients (40%) need brace and/or crutches for mobilization.

Discussion

Standard treatment for tibial hemimelia in the early Western literature was early amputation at different levels.[5,18] The main point of contention in the literature was regarding the level of amputation and requirements for reconstruction.[5,14,22] With the development of reconstruction techniques and distraction osteogenesis according to the Ilizarov principles by the mid-1990s, reconstruction became the preferred method of treatment. Gradual correction with external fixators in combination with foot and fibular centralization ensures good results.[5] We prefer to centralize the ankle joint at an early age using Ilizarov's distraction principles.

Epps et al. reported the results of fibular centralization to the posterior facet in type I tibial hemimelia with a high rate of flexion contracture at the knee.[5,14,17] We observed similar knee flexion contracture related to frame when physiotherapy is not sufficiently completed.

Simmons et al. suggested that quadriceps function with extension of the knee is mandatory to achieve satisfactory results of fibular centralization. The SF-36® (QualityMetric Inc., Lincoln, RI, USA) health survey showed that disarticulation was significantly higher than reconstruction in type I hemimelia patients (p<0.01). According to our physical and psychological findings, we suggest early amputation for type IA tibial hemimelia.

Extensive knee flexion deformities and knee subluxations are common in tibial hemimelias, as in other congenital lower extremity deformities.[23] Three knee dislocations and 2 subluxations occurred during the lengthening procedures. Decrease in knee joint motion should alert the physician to subluxation. We suggest extending the frame construction until the supracondylar region of the femur and passing the knee joint with an external hinge to prevent subluxation.

Goals of treatment are plantigrade foot, stable and functioning knee joint, stable ankle joint mostly with arthrodesis, and leg length equality. Distraction osteogenesis provides not only gradual correction, centralization of the foot, and single bone alignment of the lower extremity with tibia fibular fusion, but it also allows lengthening of the extremity.

With the exception of type IA tibial hemimelia, the primary aim of treatment should be the centralization of the foot into the posterior facet of the calcaneus in a slightly equinus position. This will permit the use the Chopart and Lisfranc joints to obtain a smoother step with a circular type external fixator.[24] It is a difficult technique which requires 3-dimensional imaging. This technique is especially preferred in type IB and type II patients with deformities which occur during childhood growth. In addition, LLD is considered for corrections during and at the end of childhood growth. Lengthening via extension the fibula before it becomes as thick as the proximal part (tibia) of the bone is not suggested. We prefer lengthening the extremity when the fibula becomes as thick as the proximal tibia.

Disarticulated patients are satisfied with their prostheses. Statistical analyses show that disarticulation is not superior to reconstruction ($p=0.63$) except in type IA patients. Our analysis showed that reconstruction in type IA was significantly poorer in comparison to disarticulation ($p<0.01$).

This study has some limitations, most of which are inherent to its retrospective design and heterogeneity of patients. The SF-10™ questionnaire has not been validated in the patient demographic included in this study. Although the mean follow-up was 5.8±3.7 years, only 6 patients had reached skeletal maturity at final follow-up.

In conclusion, reconstruction surgeries can be offered with the combination of distraction osteogenesis principles in tibial hemimelia patients. For type IA, parents and physicians should be realistic in their expectations. Disarticulation appears to be the best treatment modality when the knee is overall unstable and quadriceps function deficiency and foot deformities are present. For the other types, reconstruction surgery can provide satisfactory results. Amputation and disarticulation should be considered by parents and physicians. Reconstruction is a complex long-term treatment modality with a high rate of complications, which have a great impact on children's social life and psychology. Treatment techniques of distraction osteogenesis and Ilizarov principles should be applied by experienced surgeons in specialized centers.

Conflics of Interest: No conflicts declared.

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