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ORIGINAL ARTICLE

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Congenital longitudinal deficiency of the tibia

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Abstract We performed a clinical and radiographic review of 15 patients (19 limbs) with longitudinal deficiency of the tibia treated between 1981 and 2001. Ten limbs with Kalamchi type I deficiencies were managed by through-knee amputation. Five type II deficiencies were treated by foot ablation and tibiofibular synostosis, either at the same time or staged, but prosthetic problems may arise from varus alignment and prominence of the proximal fibula. Patients with type III deficiencies (four cases) were treated by foot ablation. Prosthetic problems relating to proximal or distal tibiofibular instability may necessitate additional surgical intervention.

Résumé Nous avons fait une révision clinique et radiographique de 15 malades (19 membres) présentant une déficience longitudinale du tibia traitée entre 1981 et 2001. Dix membres avec un déficit type Kalamchi type I ont été traités par une désarticulation du genou. Cinq type II ont été traités par ablation du pied et synostose tibiofibulaire, en un ou plusieurs temps, mais, dans ces cas des problèmes prothétiques peuvent survenir à cause d'un varus ou de la proéminence du péroné proximal. Les malades avec un type III (quatre cas) a été traité par l'ablation du pied. Les problèmes prothétiques en relation avec une instabilité tibiofibulaire proximale ou distale peuvent nécessiter une intervention chirurgicale supplémentaire.

Introduction

Congenital longitudinal deficiency of the tibia is a rare anomaly involving complete or partial absence of the tibia. Associated anatomic abnormalities identified by dissection include an abnormal vascular supply to the lower leg, dysplasia or absence of muscles, a fibulocalcaneotalar articulation rather than a true ankle joint, and coalitions between two or more tarsal bones [23]. Clinical concerns include instability (absence of normal articulation) at the knee and ankle, equinovarus foot deformity (often associated with longitudinal deficiencies), and a leg-length discrepancy that may be reach 18–20 cm by skeletal maturity.

The goal of this retrospective clinical and radiographic study was to review our experience treating 15 patients (19 limbs) over a 20-year period and to compare our findings with those of previous investigations.

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Material and methods

A review of our database from 1981 to 2001 identified 15 patients (19 limbs) with congenital longitudinal deficiency of the tibia. A chart review was done to document the physical findings, coexisting abnormalities (visceral or musculoskeletal), treatment, and clinical course. Available radiographs of the hips and lower extremities were reviewed to assess femoral morphology (length, width, epiphyseal anatomy), fibular alignment and position, and the alignment and osseous anatomy of the foot. Institutional review board approval was obtained.

Patients were classified according to the system of Kalamchi and Dawe [14]. In type I there is complete absence of the tibia, and the extensor mechanism is absent. Type II deficiencies involve the distal tibia, and patients have a well-formed proximal tibia and a functional quadriceps mechanism (Fig. 1a, b). The proximal tibia may be unossified at the time of initial evaluation, and either an ultrasound [10] or MRI may be required to differentiate type I from type II. Type III is a distal deficiency associated with diastasis of the distal tibiofibular articulation (Fig. 2a, b).

Results

Nineteen limbs in 15 patients were treated during this 20-year period. The mean age at presentation was 22 (2–55) months, and the mean follow-up was 84 (13–195) months. There were ten males and five females with ten type I, five type II, and four type III deficiencies. Four patients had bilateral involvement, three of whom had a type I deficiency on both sides, and one patient had both

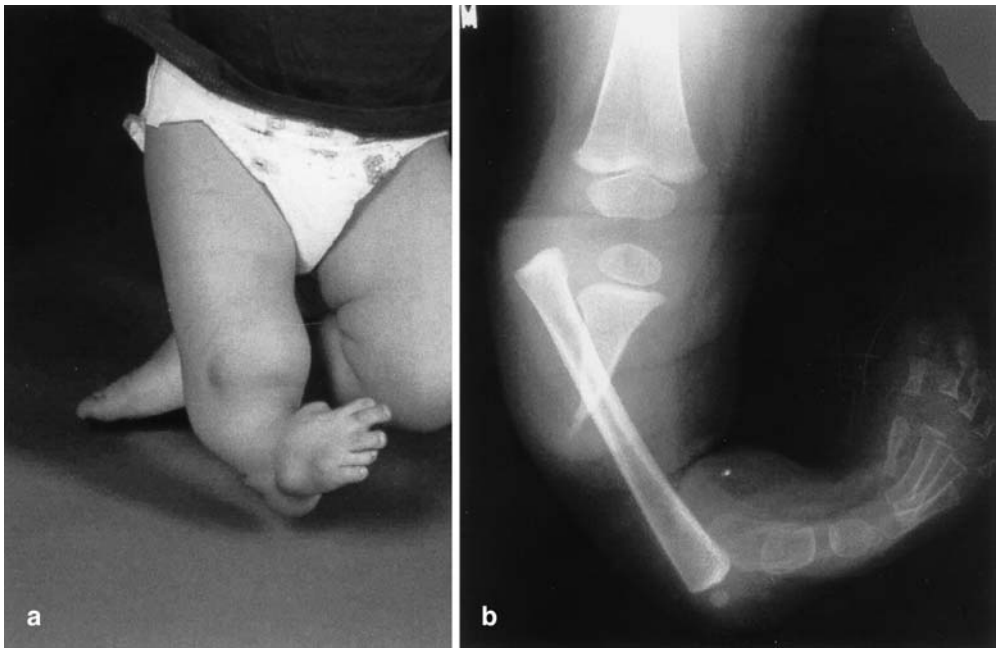

Fig. 1 Type II tibial deficiency. **a** Clinical photograph showing severe shortening associated with varus deformity. The foot is in equinovarus, with coexisting preaxial polydactyly. **b** The proximal tibia is well formed, and the lower leg is in varus despite the absence of fibular angulation. The fibular head is dislocated and has migrated proximally

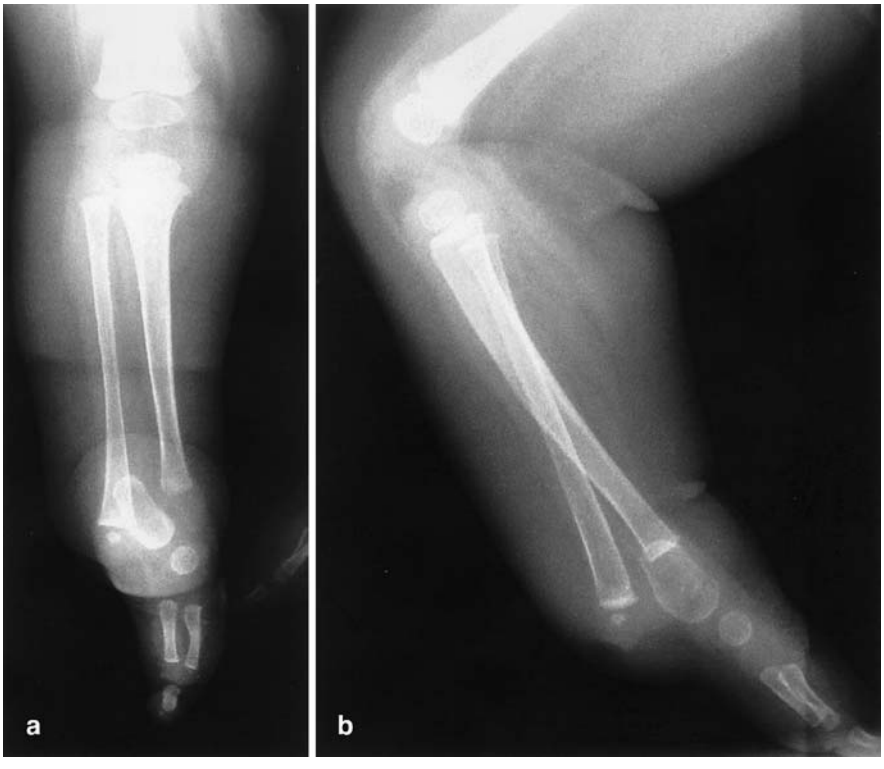

Fig. 2 Type III tibial deficiency. **a** The AP radiograph demonstrates a distal divergence, associated with a longitudinal deficiency of the foot. **b** A lateral radiograph in the same patient shows a severe equinus deformity. No proximal tibiofibular instability was identified

a type II and a ype III deficiency. Fifteen deficiencies were right sided, including all unilateral cases. Clinical findings are listed in Table 1, and radiographic findings are shown in Table 2.

Coexisting musculoskeletal (8/15) and visceral (2/15) abnormalities were observed, most commonly a congenital anomaly of the hand (Table 1).

Radiographically, type I deficiencies were associated with hypoplasia of the femur, and two patients had a distal femoral duplication (bifid condyle). The femora were symmetric in length and width in the type II and type III deficiencies, except for one case of congenital short femur and coxa vara. Proximal and posterior migration of the fibula was universal. All type II deficiencies but only one patient with a type I or type III deficiency had significant angulation of the fibula. Type III deficiencies had diastasis of the distal tibiofibular articulation, with or without proximal tibiofibular instability. All but one

Table 1 Clinical findings. *KD* knee disarticulation, *PAPVR* partial anomalous pulmonary venous return

Case	Classification/ Side (Kalamchi [14])	Coexisting visceral/ musculoskeletal findings	Primary treatment	Age	Secondary treatment	Other procedures	Complications
1	I R	None	KD	3+8	None	None	None
2	I R	None	KD	5 mos	None	None	None
3	I R	Imperforate anus, rectovaginal fistula, PAPVR	KD	2+0	None	None	None
4	I R	B central hand deficiency	KD	8 mos	None	None	None
5	I R	Femoral duplication	KD	7 mos	None	Excision medial limb bifid. femur	None
6	I R	Preaxial polydactyly (L)	KD	4+8	None	None	None
7	I R	Femoral duplication, B cleft hand	KD	3+3	None	Excision medial limb bifid. femur	None
	I R	–	KD	3+3	–	–	–
8	I R	None	KD	10 mos	None	None	None
	I L	–	KD	–	–	–-	–
9	II R	B hand anomalies (multiple)	Syme synostosis	12 mos	None	Proximal fibular epiphysiodesis, tibial stapling	Varus, prominent fibular head
10	II R	Preaxial polydactyly	Chopart	13 mos	Synostosis	None	Fibular reossification
11	II R	Scoliosis	Syme synostosis	8 mos	Synostosis	None	None
12	II R	None	Chopart	2+2	Synostosis	Debridement	Wound breakdown
13	II R	None	Syme synostosis	3+4	None	None	None
	III L	–	Syme synostosis	3+4	None	Excision of proximal fibula, synostosis	Proximal tibiofibular instability
14	III R	None	Chopart	1+5	None	None	Distal tibiofibular instability
	III L	–	Chopart	1+5	None	None	–
15	III R	Syndactyly	Syme synostosis	1+2	None	None	None

patient had an equinovarus deformity, and longitudinal deficiencies of the foot were seen in two of nine type I, one of five type II, and three of four type III deficiencies.

Patients with type I deficiencies were treated by knee disarticulation. There were no perioperative complications, and no additional surgical procedures were required. No specific prosthetic problems were identified at follow-up.

Type II deficiencies were treated initially by foot ablation (Syme or Chopart), and a tibiofibular synostosis. All patients treated initially by foot ablation alone developed prosthetic irritation in the region of the proximal fibula due to varus alignment of the lower limb associated with a prominent and unstable proximal fibula. One patient had difficulties with prosthetic fit following synostosis attributable to a progressive varus deformity. This was treated effectively by fibular epiphysiodesis and medial tibial physeal stapling 8 years after synostosis. There were no ongoing prosthetic problems at the time of the most recent follow-up.

Limbs with type III deficiency were treated by Syme amputation, and two developed complications, including symptomatic instability at either the proximal or distal articulation.

Discussion

Congenital longitudinal deficiency of the tibia represents a spectrum from partial to complete absence of the tibia. Principles of treatment are, in general, based upon the anatomy of each deformity.

We have treated all type I deficiencies with knee disarticulation, and none have required further surgical intervention and no prosthetic difficulties have been identified. Femoral shortening was a constant finding, and this provides space to maintain equal knee heights after prosthetic fitting. Two cases had an associated distal femoral duplication (bifurcation) [17, 23], and resection of the extra condyle is necessary to facilitate prosthetic fitting. Fibular dimelia has also been reported in association with a tibial deficiency [15].

Historically, the most controversial topic in the treatment of type I deficiencies has been whether to perform

Table 2 Radiographic findings. *NA* radiographs were either postoperative, did not effectively image that portion of the anatomy, or were unavailable

Case	Classification (Kalamchi [14])	Femur	Tibia	Fibula	Foot
1	I	Hypoplastic	Absent	No angulation	4 rays
2	I	Hypoplastic	Absent	No angulation	Normal osseous anatomy
3	I	Hypoplastic	NA	NA	NA
4	I	Hypoplastic	NA	NA	NA
5	I	Hypoplastic, bifid.	Absent	No angulation	2 rays
6	I	Hypoplastic	Absent	Varus (25°) and posterior (52°) angulation	Preaxial polydactyly (bilateral)
7	I	Hypoplastic, bifid.	Absent	No angulation	2 rays
	I	Hypoplastic	Absent	No angulation	2 rays
8	I	Hypoplastic	Absent	No angulation	Normal osseous anatomy
	I	Hypoplastic	Absent	No angulation	Normal osseous anatomy
9	II	Symmetric	Tapered, 50% of fibular length	Varus angulation (37°)	Hypoplastic 1st metatarsal
10	II	Symmetric	Tapered, 50% of fibular length	Posterior angulation (48°)	Normal osseous anatomy
11	II	Congenital short femur	Tapered, 50% of fibular length	Varus (27°) and posterior (11°) angulation	NA
12	II	Symmetric	Large cartilaginous mass proximally, small ossific. body	Varus (26°) and posterior (38°) angulation	Normal osseous anatomy
13	II	Symmetric	Tapered, 50% of fibular length	No lateral radiograph, fibular metaphysis above tibial physis, no AP angulation	3 rays
	III	Symmetric	Shorter than fibula	No lateral radiograph, fibular metaphysis above tibial physis	3 rays
14	III	Symmetric	Tapered, shortened	Valgus distally, proximal fibula at level of tibial physis (R)	2 rays
	III	Symmetric	Tapered, shortened	Proximal fibula 3–4 mm above the tibial physis (L)	2 rays
15	III	Symmetric	Distal segment dysplastic (tapered) with varus (32°) angulation, tibia longer than fibula	Proximal fibula posterior, metaphysis above tibial physis, no significant angulation	Normal osscous anatomy

fibular centralization (Brown procedure) [1, 2, 4, 7, 8, 11, 20] or knee disarticulation.[9, 13, 18, 19, 21]. The status of the extensor mechanism is critical to the decision making, as patients with insufficient quadriceps strength often develop disabling flexion contractures following centralization [8, 9, 14, 16, 19]. Several authors have reported favorable results in the presence of adequate quadriceps function [4, 11, 20]; however, the current consensus favors through-knee amputation, given the anticipated function with modern prostheses.

The most common approach in patients with a type II deficiency has been foot ablation and proximal tibiofibular synostosis. While amputation addresses the foot deformity and the absence of a suitable articulation at the ankle, tibiofibular synostosis realigns the lower limb and stabilizes the fibular segment [13, 14, 18, 19, 21]. Options for foot ablation include the Syme [1, 2, 9, 13, 19], Chopart [19], and modified Boyd amputations [14, 21]. The current trend in the literature is to perform a Syme or modified Boyd amputation. The foot ablation and synostosis may be either performed at the same time or staged, but we prefer to perform both procedures at the same time. One exception may be in cases in which the proximal tibia is not yet ossified, as nonunion may be more likely when the fibula is synostosed to cartilage rather than to bone [13].

Chronic skin irritation over the lateral knee during prosthetic use may be observed in those limbs treated by foot ablation alone, and we also observed this following synostosis as well. Likely causes include varus alignment of the limb, dynamic varus alignment during stance relating to lateral ligamentous instability, and prominence of the dislocated proximal fibula. Following synostosis, potential causes include the failure to accurately align the limb at the time of surgery, progressive varus deformity during growth, or prominence of the fibula. Our case of progressive varus was managed effectively with proximal fibular epiphysiodesis and medial physeal stapling. Pattison and Fixsen have documented progressive angular deformity of the fibula and have recommended treatment by osteotomy [18].

With respect to management of the proximal fibula at the time of synostosis, no guidelines are available in the literature, and we are unable to draw firm conclusions based upon this series. Options for treating a *prominent* fibula at the initial procedure include excision or epiphysiodesis; alternatively, either of these may be performed for salvage should prosthetic problems occur later.

Several different techniques for synostosis have been reported, most commonly side-to-side opposition with screw fixation. Depending on the local anatomy, Sulemaa and Ryoppy employed side-to-side opposition, implantation of the fibular segment into the tibial metaphysis, or implantation of a tapered distal tibia into the fibula [21]. In the bifurcation synostosis procedure, continuity of the periosteal tube is maintained, as the distal segment is translated medially and opposed to the lateral tibial metaphysis [5]. When reossification is complete, a Y-shaped synostosis results. Aligning the limb in neutral to slight valgus at the time of synostosis is perhaps more important than the particular technique employed. Any deformity of the fibular shaft should be taken into account when selecting the location of the fibular osteotomy. It remains possible that two osteotomies may be required if there is significant fibular angulation below the site for synostosis.

Although most authors recommend foot ablation and tibiofibular synostosis for type II deficiencies, reconstruction is technically feasible [3, 6, 12]. De Sanctis et al. used a staged approach in three patients [6]. The equinovarus foot was treated by serial casting followed by posteromedial release, and at 1–5 years of age, a proximal tibiofibular synostosis was performed. Talofibular arthrodesis was done at 3–6 years of age, and lengthening of the fibula was completed at 5–8 years of age. Javid et al. described a case in which proximal tibiofibular synostosis was followed by centralization of the fibula on the talus [12]. Realignment of the foot and fibular lengthening (15 cm.) were performed at 8 years of age. Patients treated by these approaches must be committed to multiple procedures over many years in order to preserve their foot, and it remains to be determined whether limb function and patient satisfaction will be greater in comparison with prosthetic reconstruction.

Type III deficiencies are notable for divergence and instability at the distal tibiofibular articulation, in addition to deformity of the foot with or without a longitudinal deficiency. Soft-tissue reconstruction (creation of a neoarthrosis) has been reported [22], as has talectomy and closure of the diastasis [9]. However, the majority of patients have been treated by foot ablation based upon the projected leg-length discrepancy [13, 14, 18, 19]. Schoenecker et al. reconstructed the ankle and retained the foot in nine of ten patients; however, five of nine ultimately elected to have a Syme amputation [19]. After foot ablation, two of our four limbs had prosthetic problems, attributable to either proximal or distal tibiofibular instability. The proximal instability was addressed by excision of the fibular head and proximal tibiofibular synostosis.

Finally, all 11 of our unilateral cases involved the right lower extremity. Of 215 cases in the literature [1, 4, 7, 9, 11, 13, 14, 16, 19, 22], 136 (63%) were observed on the right. In the 130 unilateral cases, 93 (72%) were right sided. The reason for this disparity remains unclear.

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