Review Article

Congenital Tibial Deficiency

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Abstract

Congenital tibial deficiency is a rare condition characterized by partial to complete absence of the tibia, an intact but frequently overgrown fibula, variable degrees of knee deformity and function, and an abnormal equinovarus foot. It can occur in isolation but also presents concurrently with other orthopaedic anomalies and syndromic conditions. Among these, congenital abnormalities of the hand and femur are most commonly observed. Many theories exist regarding its etiology and some genetic mutations have been identified; however, the underlying mechanism remains unknown. The prognosis and treatment differ based on the clinical severity. The goal of treatment is always to create a stable, functional limb, most commonly with amputation and use of prosthetics. Controversy exists over the level of amputation and the usefulness of reconstructive procedures to preserve the foot and limb length. Current investigation on this complex disorder is focused on identifying its origins and further developing a classification-based treatment algorithm to improve patient outcomes.

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Congenital tibial deficiency occurs in 1 in 1 million live births.[1] It exhibits a spectrum of disease, with varying amounts of tibia absence contributing to a range of associated knee and foot abnormalities. Knee instability is frequently present, with the most severe forms of deficiency lacking a patella and a knee extensor mechanism. Very severe forms also often have a skin dimple overlying the proximal tibia and accompanying knee flexion contracture. In less severe cases, the proximal tibia and knee extensor mechanism are present and the knee is stable. The fibula is typically intact, and the foot position varies in relation to the knee, with supination and rigid equinovarus of the foot most commonly observed.[1-5]

Embryology

In normal embryologic development, limb bud orientation progresses under three influences: the apical ectodermal ridge, the zone of polarizing activity (ZPA), and the Wnt signaling pathway. Cell-mediated interactions among these centers occur between the 4th and 7th week of gestation. The apical ectodermal ridge modulates the proximal to distal development. The ZPA produces sonic hedgehog (Shh) protein, which directs anterior versus posterior orientation; this becomes radioulnar in the upper limb and the tibia and fibula in the lower limb. The dorsal limb bud secretes Wnt protein, which distinguishes dorsoventral development.[6]

Most limb deficiencies are present by the 7th week of gestation. The upper limb develops after day 28 and the lower limb by day 31, and both progress proximal to distal. Upper and lower limb abnormalities appear after days 35 and 37, respectively. Although the exact cause and timing of tibial deficiency remain unknown, the responsible insult likely occurs during this phase.[7]

One theory for the cause of limb deficiency is vascular insufficiency. Levinsohn et al[8] reported on tibial and fibular deficiency and clubfoot. In all cases, they found similar patterns of arterial deficiencies, involving the anterior tibial and dorsalis pedis artery. Vascular insufficiency can create either pre- or post-specification defects, depending on the timing in relation to mesenchymal differentiation. Post-specification defects leave a rudimentary structure, which, the authors postulate, explains tibial deficiency given the presence of a partially formed proximal tibia. However, others speculate a pre-specification cause, because the limb bud starts off in close proximity to the mesenchyme of other vital organs, providing an explanation for the coexistence of other systemic developmental anomalies.[3]

Table 1

Percentage of Observed Associated Pathology in the Two Largest Series of Congenital Tibial Deficiency

Associated Pathologies	Schoenecker et al[2] (57 pts) (%)	Clinton and Birch[5] (95 pts) (%)
Overall	60	78
Bilateral	25	32
Upper extremity	30	33
Cleft hand	9	16
Radial deficiency	1	8
Other	20	9
Lower extremity	48	96
Deficient lateral rays	9	21
Medial ray/great toe duplication	—	14
Hip dysplasia or dislocation	18	11
Congenital femoral deficiency	9	11
Coxa valga	12	—
Other	—	40
Visceral	NA[a]	43
Cardiac	—	21
Gastrointestinal	—	15
Genitourinary	—	7
Spine	21	12

[a] Visceral organ involvement not reported by Schoenecker et al.

Genetic Basis

No specific gene mutation has been identified as the cause of congenital tibial deficiency. The Shh pathway has been implicated in syndromic forms.[9] Recent research has identified a 5 kb deletion within the DNA of the Shh repressor Gli3 protein in two patients with bilateral tibial deficiency, resulting in unrestricted Shh activity outside the ZPA.[10] This study also noted that the genetic deletion occurred only on the maternal allele, suggesting autosomal-dominant inheritance with incomplete penetrance.[10] However, both autosomal-dominant and autosomal-recessive inheritance models have been described,[11-13] and case reports of additional chromosomal abnormalities exist.[14] There are likely multiple genetic alterations and inheritance patterns responsible for tibial deficiency.

Syndromic forms of tibial deficiency also exist, with four known associated autosomal-dominant types: Warner's Syndrome, tibial hemimelia diplopodia syndrome, tibial hemimelia–split hand and foot syndrome, and tibial hemimelia-micromelia-trigonobrachycephaly syndrome. Among these disorders, a wide range of clinical expression exists, supporting variable penetrance. For example, in a series of 37 patients with tibial hemimelia–split hand and foot syndrome, severity ranged from isolated digit hypoplasia or syndactyly to complete bilateral tibia agenesis with split hands.[15]

Associated Pathology

Other congenital abnormalities are observed with high frequency. Congenital hip dislocation, vertebral malformations, bifurcation of the femur, imperforate anus, and hypospadias have been reported.[2,3,16] In an older series of 57 patients studied by Schoenecker et al,[2] 34 patients (60%) had an associated abnormality of the hip, hand, or spine. The most common hip abnormality was congenital dislocation (10 patients), followed by coxa valga and proximal focal femoral deficiency. Hand deformities were also prevalent, occurring in 17 patients (30%). Spine abnormalities were present in 12 patients (21%), including 5 with hemivertebrae, as well as hypoplastic vertebrae, scoliosis, and spina bifida. Interestingly, 20 patients (35%) reported a family history of congenital anomalies ranging from hand deformities (10 patients) to congenital tibial deficiency (5 patients). Most of these were in first-degree relatives.

A more recent study by Clinton and Birch[5] details a longitudinal series of 95 patients spanning 37 years at Texas Scottish Rite Hospital. Among these patients, 79% had other abnormalities, consistent with previous reports. These were mostly other lower extremity anomalies,

Table 2

Number of Observed Additional Upper Extremity, Lower Extremity, and Spine Abnormalities, Organized by Jones Type in 71 Limbs Described by Schoenecker et al[2]

Type	Upper Extremity, No. (%)	Lower Extremity, No. (%)	Spine, No. (%)	Total Limbs
1A	6 (18)	12 (36)	5 (15)	33
1B	1 (17)	3 (50)	2 (33)	6
2	2 (12)	6 (37)	3 (19)	16
3	3 (43)	1 (14)	1 (14)	7
4	4 (44)	1 (11)	1 (11)	9
Totals	16 (23)	22 (31)	12 (17)	71

Table 3

Number of Observed Additional Upper Extremity, Lower Extremity, Visceral Organ, and Spine Abnormalities, Organized by Jones Type in 125 Extremities Described by Clinton and Birch[5]

Type	Upper Extremity, No. (%)	Lower Extremity, No. (%)	Visceral, No. (%)	Spine, No. (%)	Total Limbs
1A	23 (32)	44 (60)	23 (32)	12 (16)	73
1B	1 (17)	1 (17)	1 (17)	1 (17)	6
2	6 (33)	11 (61)	5 (28)	4 (22)	18
3	1 (50)	2 (100)	0 (0)	0 (0)	2
4	4 (33)	5 (42)	2 (17)	0 (0)	12
5	2 (14)	12 (86)	2 (14)	1 (7)	14
Totals	37 (30)	75 (60)	33 (26)	18 (14)	125

such as absent lateral rays and hip dislocation. Upper extremity anomalies, such as cleft hand, radial deficiency, and congenital scoliosis, also occurred. Congenital tibial deficiency, unlike other longitudinal deficiencies, is particularly notable for associated visceral organ involvement. This series noted that 20 patients had an associated cardiac malformation, 14 had a gastrointestinal malformation, and 7 had a genitourinary congenital abnormality. These two studies are the largest reports of observed congenital anomalies (Tables 1–3).

Classification Systems

The Jones classification is the most commonly used system, dividing tibial deficiency into four groups, from "worst" to "best," based on the skeletal morphology of radiographs of affected infants.[17] Type 1, which lacks any tibial ossification, has two distinct clinically relevant subgroups. Type 1b contains a cartilaginous anlage, whereas type 1a has no cartilagenous anlage and further has a hypoplastic distal femoral epiphysis. Type 2 demonstrates ossification of the proximal tibia. In type 3, the least common form, isolated ossification of the distal tibia is seen. Type 4 is short tibia with an absent distal articular surface and distal tibiofibular diastasis (Figures 1 and 2). Although this classification scheme remains the most widely adopted, limitations exist owing to the broad clinical spectrum of the condition.

Kalamchi and Dawe[18] and Weber[19] modified the Jones classification based on their experience. They simplified the Jones classification into three groups, omitting Jones type 3 patients because this form was never encountered at their institution. Arguing that 15% of patients could not be described according to the Jones method, Weber created a more elaborate classification, incorporating seven categories from "best" to "worst" and 5 subcategories based on the presence or absence of a tibia cartilaginous anlage, which affects reconstruction. Type 1 is hypoplastic, type 2 is distal diastasis, type 3 is distal aplasia, type 4 is proximal aplasia, type 5 is bifocal aplasia, type 6 is agenesis with a double fibula, and type 7 is agenesis with a single fibula (Figure 3).

A final classification system, first proposed by Paley[21] in 2003 and later modified, describes both the progressive spectrum of deficiency and the treatment algorithm for each type[20,21] (Figure 4). Type 1 represents a congenitally short tibia with relative fibula overgrowth. In type 2, the proximal and distal epiphyses are present with a dysplastic ankle. The tibia plafond is absent or deficient in type 3 with distal diastasis. Only the proximal tibia is present in type 4. Type 5 represents the most severe with complete absence of the tibia (Table 4).

Epidemiology Based on the Jones Classification

Schoenecker et al[2] presented in their report a distribution of commonly encountered forms. Type 1a and 2 deficiencies were the most common, representing 46% and 21% of the series, respectively. Type 4 occurred in 14%, and type 3 and 1b were the least common, affecting 9% and 8% of limbs, respectively. In this series, no limbs were described that did not fit the Jones classification.

Consistent with Schoenecker et al, Clinton and Birch[5] reported a similar

epidemiologic pattern of tibial deficiency. Among them, type 1a was the most common, occurring in 58% of limbs. Type 2 was the second most common, occurring in 14%. No limb was truly type 3, because all eventually developed a proximal tibia epiphysis on radiographs. Fourteen of 95 limbs (11%) were characterized by more global tibial deficiency and deemed not classifiable by Jones criteria. All patients had similar radiographic appearance, with proximal and distal tibia epiphyses, and proximal and distal fibula overgrowth leading the authors to propose a distinct "Jones 5" group. Of note, a wide range of deformity coexisted with this group which affected management. Table 5 details the relative frequency of Jones types observed in these two large series.

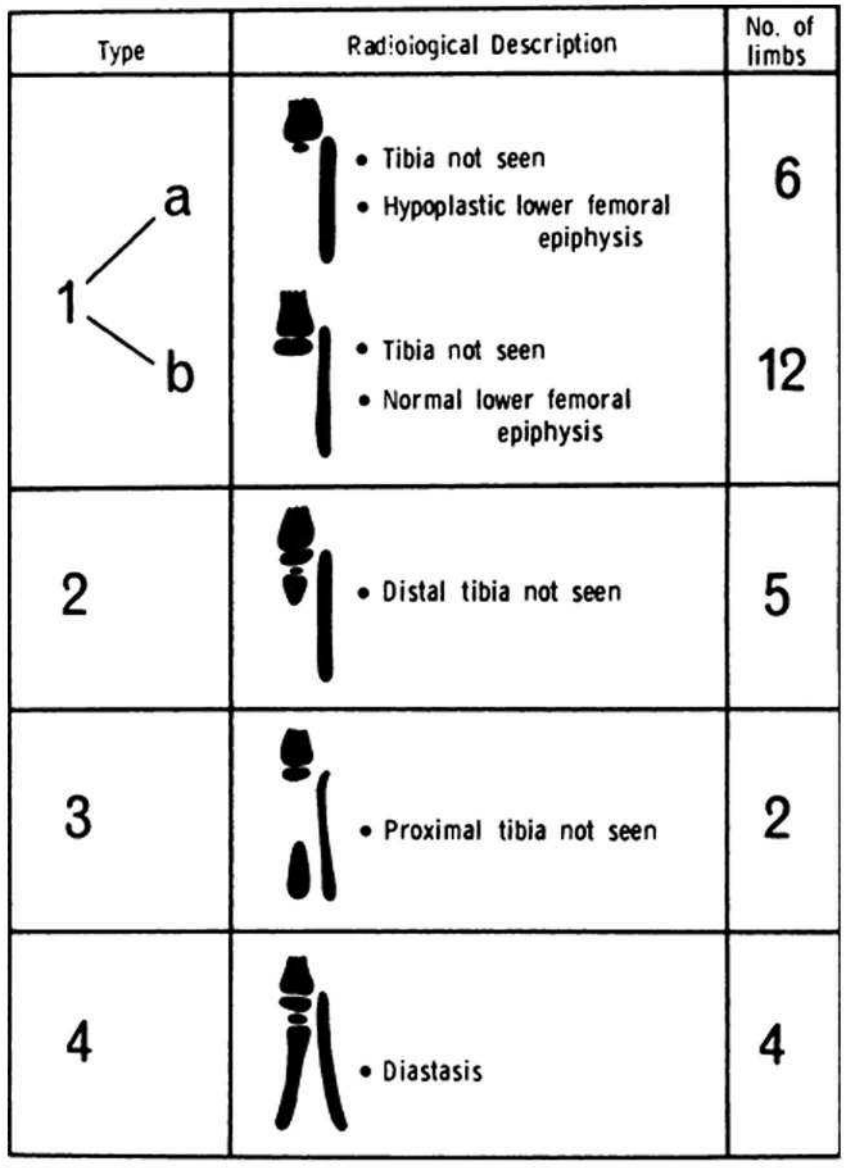
Clinical Features

A broad spectrum of clinical pathology exists in congenital tibial deficiency. Physical examination of the affected limb follows the Jones classification. In Jones type 1a, with complete tibial aplasia, hamstring function is present and quadriceps function is deficient, causing knee flexion contracture. The patella does not form, and the foot is in rigid equinovarus. In Jones type 1b and 2, the knee extensor mechanism is formed, producing a functional knee without contracture. The fibula displaces proximally and laterally, and an equinovarus foot is noted. In Jones type 3, with isolated distal tibia ossification, the knee is unstable, with varus positioning of the overall limb. Jones type 4 patients have a stable knee and a rigid equinovarus foot positioned in the diastasis between the tibia and fibula.

The equinovarus foot position akin to all types of congenital tibial deficiency can be confused with an isolated clubfoot, a common reason for initial orthopaedic referral. An atypically rigid clubfoot, absent lateral rays, or medial ray duplication should alert the orthopaedic surgeon to investigate for tibial deficiency. We stress the importance of fully evaluating the entire lower extremity for clues that the equinovarus foot may be a manifestation of a longitudinal deficiency, because the treatment and prognosis are vastly different (Figure 5).

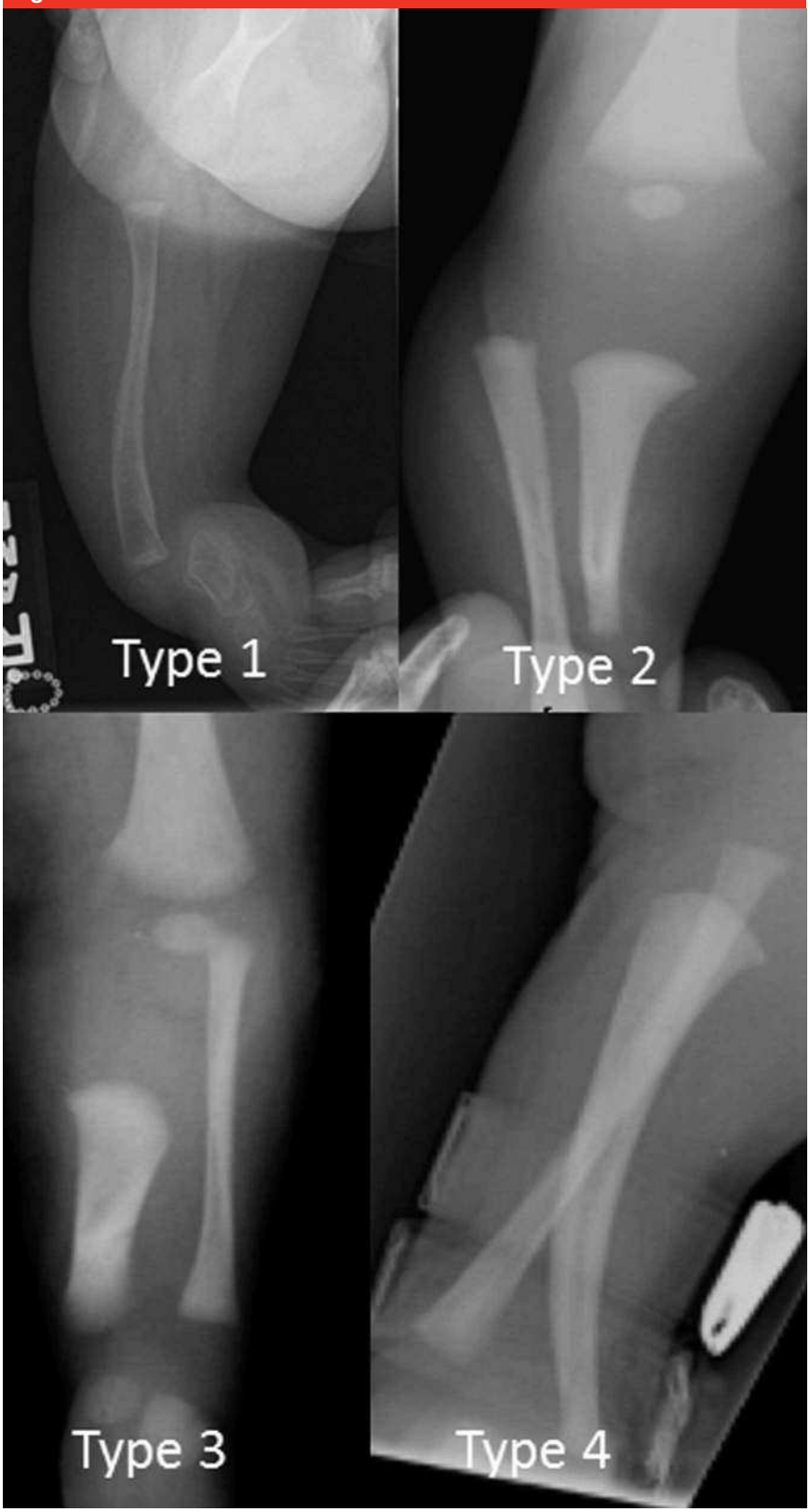
In addition to the characteristic clinical findings, congenital tibial deficiency is distinctive for frequent associated congenital abnormalities, which do not correlate well with any classification system. Other orthopaedic anomalies are commonly encountered in all forms, and a thorough

Figure 1



Jones classification of congenital tibial deficiency. (Reproduced with permission from 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;60:31-39.)

Figure 2



Radiographic examples of each Jones type of congenital tibial deficiency.

physical examination of all extremities, hips, and the spine is essential with a low threshold to obtain additional imaging. Because of the risk of associated visceral organ abnormalities, we recommend a genetics consultation and advanced imaging to evaluate for other organ dysfunction.

Surgical Intervention

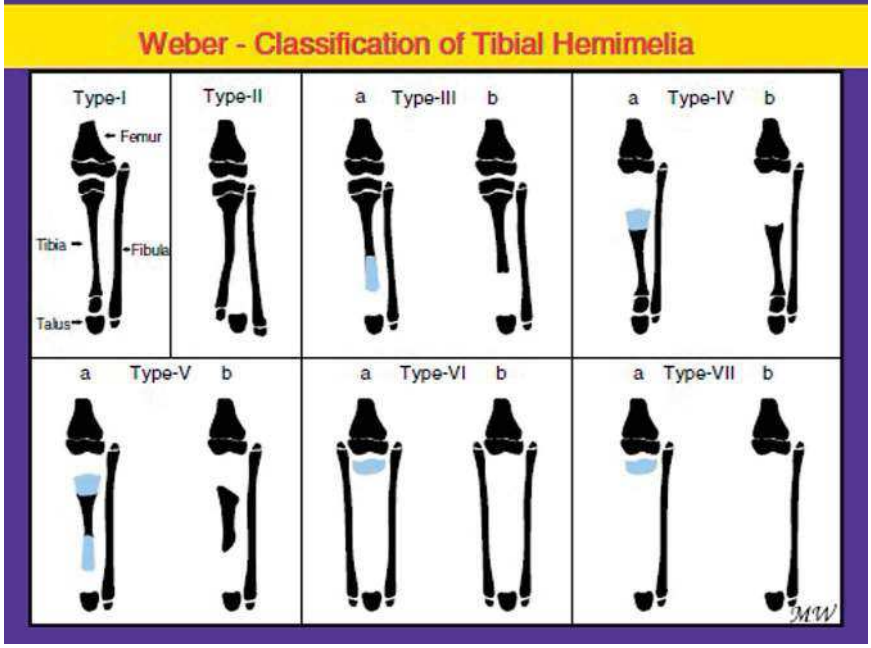
The most fundamental treatment principle is to determine whether the knee is stable with a functional extensor mechanism. Type 1a deficiencies lack any tibia. Hamstring function but not quad function is present, creating a nonfunctional contracted and displaced knee joint proximal and lateral to the femoral condyles. For this reason, the standard management of Jones type 1a tibia deficiency is knee disarticulation. In all other types of tibial deficiency, with a theoretically functional knee, an attempt is made to reconstruct the proximal tibia and fibula and preserve the knee joint. Because of the severity of foot and ankle deformity and instability, the distal limb is often managed with a Syme amputation and prosthetic fitting. For Jones type 1b and 2 limbs in which varying ossification of the proximal tibia is present, traditional management consists of proximal tibiofibular synostosis and distal Syme amputation.

It is vitally important to differentiate type 1a and 1b deficiencies because these types distinguish a nonfunctional and functional knee and extensor mechanism. Though less commonly encountered, in type 1b deficiency, the cartilaginous anlage of the proximal tibia will ossify, allowing the knee joint to be preserved. Ultrasonography is a simple method that can identify a cartilaginous tibial anlage predictive of future ossification. Additionally, ultrasonography elucidates the presence of other

important components of knee stability, including an intact patella tendon and functioning quadriceps mechanism.[22] Advanced imaging with MRI provides more precise detail, and in some cases, it may be helpful in determining whether knee reconstruction options are possible and aid in surgical preparation.[23]

The rare Jones type 3 deficiencies are commonly managed with a Syme or Chopart amputation, assuming eventual ossification of a proximal tibial cartilaginous anlage and a functional quadriceps mechanism. Similarly, the modified Syme ankle disarticulation has been the standard management of type 4 deficiencies associated with distal diastasis. Alternatively, lengthening and reconstruction options that reposition and achieve a plantigrade foot may be possible. Foot preservation techniques are often hindered by the absent distal tibia and notable deformities of the talus and calcaneus.

Figure 3



Weber classification of congenital tibial deficiency. (Reproduced with permission from Weber M: New classification and score for tibial hemimelia. *J Child Orthop* 2008;2:169-175.)

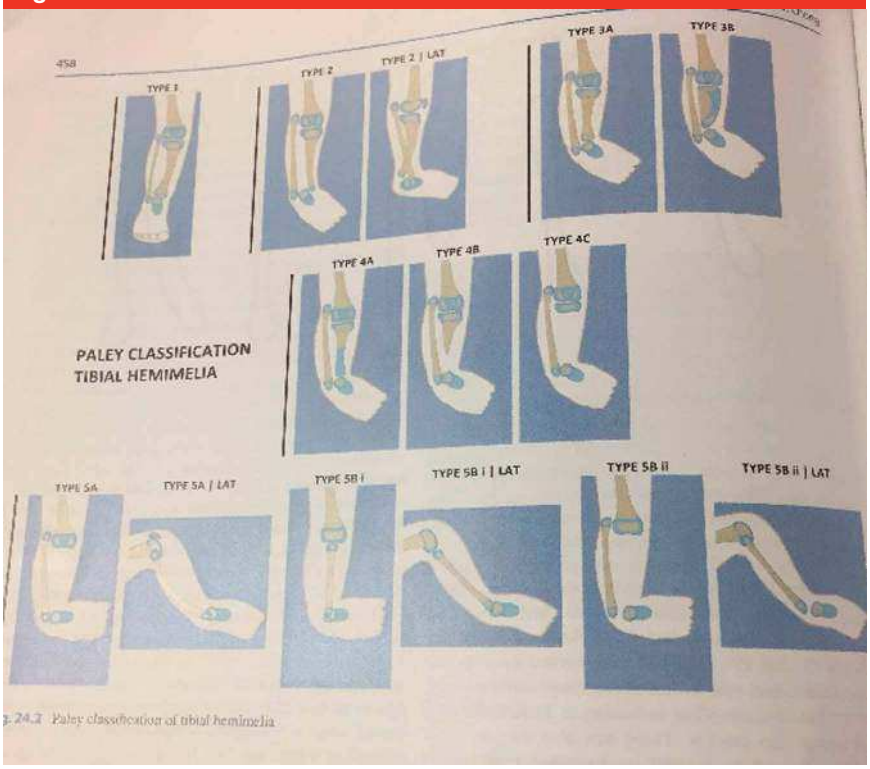
Brown Procedure

In 1965, Brown[24] described a fibular centralization procedure for congenital tibial deficiency. In this procedure, a U-shaped incision was made at the level of the knee joint and the fibula was dissected from the surrounding soft tissues through a lateral parapatellar arthrotomy. The proximal 3/8 inch of the fibular epiphysis was osteotomized to make a flat surface and was then centralized and fixed with K-wires underneath the femoral condyles. The soft tissues were imbricated to centralize the patella and tighten the capsule; distally, the patellar tendon was reattached to the centralized fibula. This procedure was largely undertaken in Jones type 1a patients. Although initial enthusiasm was present, a 15-year follow-up study revealed that most of these patients went on to have a knee disarticulation.[25]

Other series similarly reported poor outcomes after the Brown centralization procedure. Epps et al[26] published a series of 14 patients with complete tibial deficiency treated within the first year of life. All patients developed severe flexion deformity of the knee which impaired gait and interfered with prosthetic wear, and they underwent secondary surgeries to manage either the flexion deformity or knee disarticulation. The seven patients who underwent knee disarticulation obtained a satisfactory result, whereas the others remained limited by their knee flexion contracture at final follow-up. Clinton and Birch[5] also noted a high rate of knee disarticulation after the Brown procedure and knee flexion contractures in a small number of patients who did not have further revision surgery.

Although a high rate of conversion to knee disarticulation is present, the Brown procedure has been successful in patients who meet certain indications. Christini et al[27] found that 5 of 13 patients who underwent the Brown procedure had acceptable function, despite the fact that one had a knee flexion contraction and another had a limited range of motion. Similarly, Simmons et al[28] reviewed seven patients followed an average of 7 years after Brown procedure and also documented good results. The average arc of motion was 57°, and all were ambulating with patellar tendon–bearing prostheses and thigh extensions for collateral support. Both Christini and Simmons found that the Brown procedure may lead to subjectively reported acceptable function in patients with complete tibial deficiency, as long as they had a functioning quadriceps of at least grade 3 strength preoperatively. Other important criteria for a functional outcome included the absence of fibular bowing and pterygium folds in the popliteus fossa which lead to progressive flexion contractures, and age less than 1 year so the fibula has adequate time to hypertrophy with growth.[27,28]

Figure 4



Paley classification of congenital tibial deficiency. (Reproduced with permission from Paley D: Tibia hemimelia: New classification and reconstructive options. *J Child Orthop* 2016;10:529-555.)

Notable advantages exist in selecting a more distal level of amputation to preserve the native knee joint. Patients benefit from improved energy expenditure, gait efficiency, and proprioception. Although fibular centralization appears to be unsuccessful for many patients with complete tibial deficiency, those who demonstrate some preoperative active knee function may be candidates. Identifying these patients by thorough physical examination and adjunct ultrasonography will help select appropriate patients for the Brown centralization procedure.

Knee Reconstruction

Given the mixed results of the Brown procedure, other surgical techniques have been developed to recreate a functional knee mechanism. In 2002, Weber[29] proposed a technique in which the patella anlage is transposed to articulate with the distal femur. To facilitate the transposition, the quadriceps tendon is Z-lengthened and stabilized by the creation of two visor flaps made from the surrounding capsular tissue and crossed to provide medial and lateral support. The fibula can then be centralized below the patella and attached to the patella tendon. The construct is supported with a ringed fixator, which gradually increases the range of motion of the new knee joint. In theory, this surgical technique improves on the Brown centralization by adding better knee stability through the creation of the capsular visor flaps and by using the patella to provide a larger, more stable articulating surface for the distal femur, but long-term functional outcome studies are unavailable.

Reconstruction Principles

Advances in technology, our knowledge of biology, and better understanding of the pathology of all types of tibial deficiency may lend alternative treatments to amputation in the future even for the most severe types. Certain principles apply for reconstructive surgery to successfully create a functional limb in the hands of an experienced surgeon. No single intervention can address the complexity of the limb deficiency. Multiple staged procedures to realign, recreate and stabilize the joints, and lengthen the leg must be anticipated. A plantigrade foot and stable ankle must be achieved. Reconstruction of the knee, restoration of a functional extensor mechanism, and elimination of the flexion contracture must be addressed. Repeated lengthenings of the tibia or centralized fibula may be required and the adjacent joints must be stabilized. Paley[21] elaborately described reconstruction options that correspond to his classification of tibial deficiency (Table 4). Functional outcome studies of these new and modified techniques are unavailable.

Distal Amputation Versus Reconstruction

Less controversy exists over the management of partial tibial deficiency (Jones Ib-Jones 2). A tibiofibular synostosis can be performed by first osteotomizing the fibula at the neck and fusing the distal portion of the fibula to the remnant tibia in an end-to-end or side-to-side fashion with supplemental screw or plate and screw fixation (Figure 6). Because the fibula typically is migrated proximally, the technique involves resecting the proximal fibula to

Table 4

Paley Classification and Reconstruction Principles

Paley Classification	Features	Reconstruction Options
1	Congenitally short tibia overgrown fibula Proximal valgus Normal distal plafond	Correction of valgus deformity and lengthening
2	Deficient tibia plafond with diastasis of tibia and fibula	Reconstruction of ankle joint
	Foot follows the fibula	Correction of any tibia deformity + lengthening
3a	Distal tibia physis formed but separate from proximal physis	Reconstruction of ankle joint
	Plafond dysplastic	Correction of tibia deformity + lengthening[a]
	Overgrown fibula	Fibula management:(1) resection of diaphysis to create non-union and (2) distraction of tibia without fibula fixation
3b	Delta tibia representing proximal and distal physis connected through bracket epiphysis	Excision of bracket
	Malorientation of knee and ankle	Acute correction of tibia deformity + partial resection fibula
	Overgrown fibula	Lengthening[a]
4a	Delayed ossification of tibia Absent distal physis Ankle joint present but nonfunctional Overgrown fibula	Creation of plantigrade foot with stable ankle Correction of tibia deformity Lengthening after anlage ossifies[a]
4b	Complete absence of distal tibia Overgrown fibula	Correct foot deformity Fuse talus to distal fibula Transfer fibula diaphysis to distal end of proximal tibia Future lengthening of single bone leg[a]
4c	Proximal epiphysis present but absent physis Knee joint present Notable overgrown fibula	Correct foot deformity and knee contracture Fuse talus to distal fibula Fibula fixed to tibia epiphysis Repeated future lengthenings[a]
5a	Complete absence of tibia Patella present Knee flexion contracture	Patella converted to a tibia plateau Fibula centralized to patella (Weber procedure)
5b (i)	Complete absence of tibia No patella Knee flexion contracture *Fibula autocentralized*	Correction of knee contracture Centralize foot to distal fibula Reconstruction of knee ligaments and transfer quad to fibula Fuse talus to fibula Repeated future lengthenings[a]
5b (ii)	Same as 5b (i) *Fibula dislocated*	Centralize fibula to femur Correction of knee contracture Centralize foot to distal fibula Repeated future lengthenings[a]

[a] Recommends extending the external fixator to the femur to stabilize the knee.

avoid prosthetic fit problems from the protruded fibular head. Regrowth of the resected proximal fibula may be prevented by removing the periosteum.

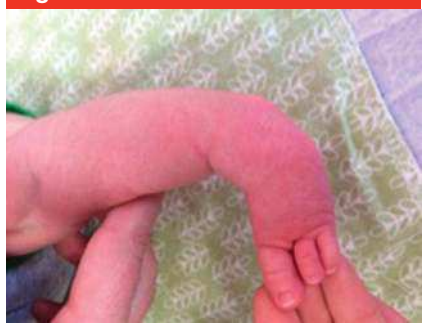
Distally, the limb is traditionally managed with either a Syme amputation, an ankle disarticulation that maintains the heel pad, or Boyd amputation, which differs because the calcaneus is retained, centralized, and fused to the proximal limb. Schoenecker et al[2] used distal Syme amputation in 12 patients with type 2 deficiency and successfully combined this with tibiofibular synostosis in 8 of them. Christini recommended the Boyd procedure instead of a Syme amputation if the calcaneus can be centralized, and some surgeons suggest that retaining the calcaneus better maintains the heel pad position.[27] The Boyd procedure is technically more difficult

Table 5

Relative Frequency of Observed Forms of Congenital Tibial Deficiency in the Two Largest Series, Organized by Jones Type

Type	Schoenecker et al[2] (71 Limbs) (%)	Clinton and Birch[5] (125 Limbs) (%)
1a	46	58
1b	8	5
2	23	14
3	10	2
4	13	10
Unclassifiable	0	11

Figure 5



Clinical photograph of an extremity affected by congenital tibial deficiency.

than the Syme, and good functional outcome is achieved through the use of a prosthetic with either amputation. However, some surgeons advocate for distal reconstruction, which depends on the length of the affected limb and the amount of deformity present in the foot. Different combinations of limb lengthening and foot centralization and reconstruction have varying success.

The first report of foot centralization was by Hosny.[30] In addition to performing fibular centralization, an Ilizarov frame was used to center the foot underneath the fibula. Since then, several series have detailed the results.[31-33] In a recent publication, Balci et al[32] presented a clear protocol for foot centralization and the results in 17 patients with partial tibial deficiency. Their protocol involved initially addressing the ankle, followed by centralization of the fibula under the remaining tibia. Achilles tenotomy and posteromedial release were performed first to mobilize the foot. Next, the posterior facet of the calcaneus was centralized under the fibula with a circular external fixation. Finally, the fibula was osteotomized and centralized beneath the tibia and held with a Steinman wire. Distraction osteogenesis for limb lengthening in a staged fashion was performed, once the distal fibula had healed to the proximal tibia and had expanded to its width.

The goals of this procedure are to obtain a plantigrade foot, a stable and functional knee joint, and a stable ankle joint with arthrodesis, as well as to equalize leg lengths. The use of distraction osteogenesis provides gradual centralization of the foot and eventual lengthening of the extremity. Although successful in achieving these goals, this approach requires multiple surgeries and has a high rate of complications. On average, patients underwent an average of 6.4 surgeries, spent 17 months in an external fixator, and experienced 5.5 complications. The severity of these complications varied, including knee subluxation or dislocation, knee flexion contracture, equinus deformity, fracture through the regenerate, revision of the circular fixator, and pin tract infections. It is also important to note that the functional outcomes after this reconstructive procedure compared with knee disarticulation in Jones 1a deficiency were not different. Furthermore, 13 of 23 patients who underwent reconstruction required bracing treatment or crutches at long-term follow-up, including all of Jones type 1 patients.[32] Finally, neither report specifically discusses the outcomes of feet with or without deficient lateral rays. It is logical to consider that the status of the foot may in part dictate the success of centralization, but this has not been reported.

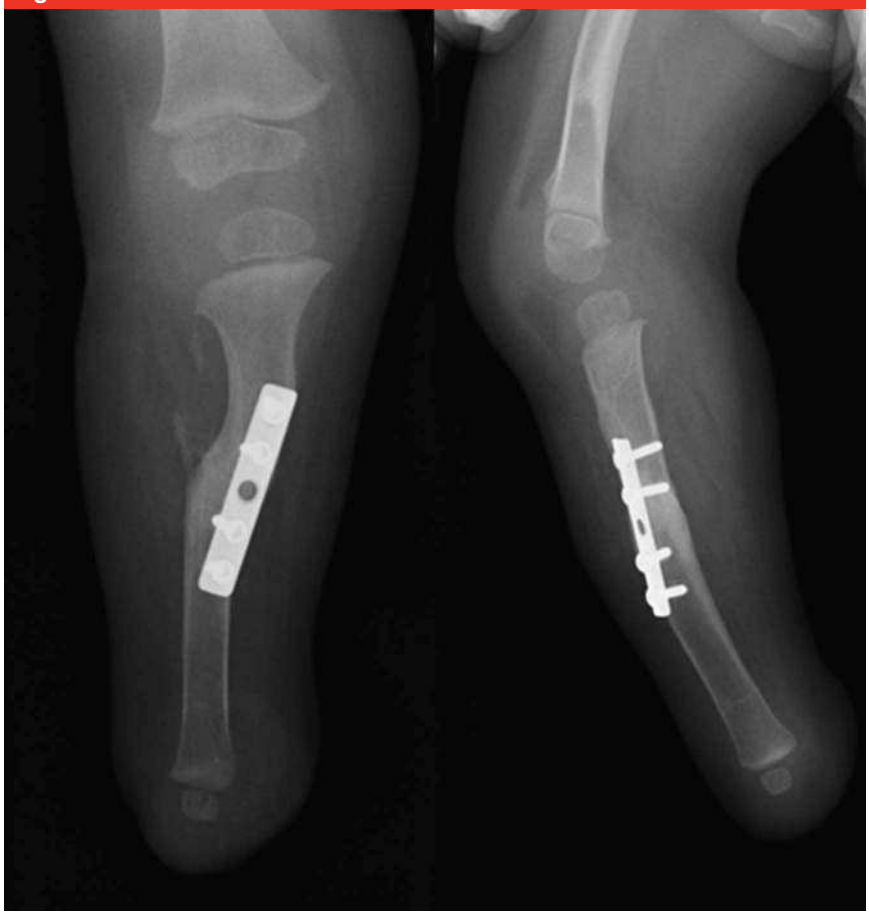
Another long-term study evaluated the outcomes between patients with primary amputation and those with distal foot reconstruction using the Pediatric Quality of Life questionnaire. Although the scores were not statistically different, in many areas patients with reconstruction scored slightly higher. However, it should be noted that the amputation group was a much smaller group of patients and had proportionately more bilateral cases than the reconstruction group. Although distal reconstructive procedures require long treatment times in a frame and high rates of complications, many patients and families select this option and report good satisfaction rates.[33]

Timing of Surgical Intervention

The optimal time to proceed with surgical intervention may be individualized. Amputation is best performed between the ages of 6 months and 1 year to allow early prosthetic fitting at the onset of walking. Patients with an additional upper extremity deficiency present an exception to this rule. Delaying lower limb amputation is appropriate until surrogate use of the lower extremity to replace the upper extremity is established. For management of partial tibia hemimelia, a

proximal tibiofibular synostosis can be performed when sufficient ossification of the upper tibia develops to permit successful union, but Jones argued that stability can be achieved with fusion to the cartilaginous anlage that will eventually ossify.[17] The surgical plan may vary considerably with multiple staged procedures anticipated to achieve a functional limb for patients suitable for more complex reconstruction procedures. Brown[24] initially recommended fibular centralization surgery by 1 year of age to maximize early ambulation, fibular articulation, and hypertrophy potential. Similarly, the Weber[29] procedure was first described in a 15-month-old child. Paley recommends achieving a stable ankle and plantigrade foot, followed by secondary procedures to create a one-bone leg and reconstruct the knee.[20,21] The timing of additional lengthening procedures is driven by the projected discrepancy, expected number of procedures, the child's functional needs, and preparedness of the family for lengthening surgery. In general, the discrepancy should be addressed with as few surgeries as possible to achieve acceptable alignment, a plantigrade foot, a stable knee, and equal limb lengths by the time of skeletal maturity. Serial lengthenings can be spaced throughout childhood to allow sufficient time without surgery. We suggest engaging the parents in a discussion of treatment options at the initial consultation and beginning reconstruction within the first few years of life.

Figure 6



Radiographic example of a patient after tibiofibular synostosis.

Psychosocial Outcomes

Children with limb deficiencies experience a high level of function. Overall, they are better equipped than the adult population to deal with a prosthetic limb. Most patients are able to wear their prosthetic for long intervals during the day. Although most of them do not have problems with pain, in one survey, 16% reported having at least moderate pain.[34] Pediatric patients with prosthetics may struggle compared with their peers. Their different appearance and physical limitations compared with typically developing children may predispose them to difficulty with psychosocial adjustment.

Michielsen et al evaluated a group of 56 Dutch children and adolescents with lower limb deficiencies. In this assessment, 8- to 18 year olds reported their participation in leisure activities and their health-related quality of life. Their general participation and health-related quality of life were not different from reference values. However, adolescents reported less diversity in the activities they participated in and less involvement in social and skill-based activities. None of these findings correlated with the degree of limb deficiency.[35]

This research does highlight the fact that children with limb deficiencies and prosthetics are a highly adaptive group who enjoy participation in leisure activities. It is notable that adolescents did struggle in a few areas. It would be worthwhile to examine the transition from childhood to adolescence in this population and better understand what contributes to their differences. Furthermore, comparisons between patients with limb reconstruction

and those with amputation may better describe how these procedures affect childhood.

Summary

Congenital tibial deficiency is an uncommon and complex condition, with a wide spectrum of clinical presentations. The main goal of treatment underlying all types is to create a stable and functional limb. The surgical options to achieve the best functional outcome vary based on the amount of deficiency present. In complete absence of the tibia, early amputation and prosthetic use provides the most reliable outcome. In the presence of a functional knee joint, reconstructive procedures and more distal amputation can be performed with good results. Surgeries that reconstruct the knee joint, such as the Brown procedure, have not been successful in most patients. However, patients who meet specific indications may benefit. Other reconstructive procedures, such as limb lengthening and foot centralization, may also be appropriate for select patients, but it is critically important that these patients and families thoroughly understand the high risk of complications, potential number of surgeries involved, and length of treatment before embarking on this path. Newer limb lengthening and surgical techniques may lead to better methods of managing the complexities of tibia deficiency in the future, making reconstruction options more attractive. Given the rarity and complexity of treating these patients, surgeons should not hesitate to coordinate care with other experienced surgeons to ensure the best technical outcome.

In addition to developing the surgical treatment plan, other important aspects are involved in the care of patients with congenital deficiency. Owing to its known association with other syndromes and organ dysfunction, it is important for each patient to be thoroughly evaluated for coexisting congenital anomalies. Additionally, more clearly understanding the psychosocial needs of affected children may in turn allow healthcare professionals to provide better support and adaptive strategies for patients and families. Finally, further research aimed to determine the underlying etiology may help identify and counsel at-risk families in the prenatal period.

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