Congenital Tibial Deficiency: A 37-Year Experience at 1 Institution

Rebecca Clinton, MD and John G. Birch, MD, FRCS(C)

Background: The purpose of this study is to evaluate all cases of tibial deficiency seen at a single institution from 1975 to 2012, to classify these cases by the Jones classification if possible, to evaluate for associated anomalies, and to review the surgical treatments provided to these patients.

Methods: Ninety-five patients (125 extremities) treated at our institution between 1975 and 2012 with tibial deficiency had complete records allowing for classification and review of full treatment course. These patients’ records and imaging were retrospectively reviewed for any associated anomalies, surgical treatment performed, and limb deformity characterized by the Jones classification where possible.

Results: Seventy-three of 125 limbs (58%) were classified as Jones type 1A, 6 (5%) as type 1B, 18 (14%) as type 2, and 12 (10%) as type 4. Two limbs initially classified radiographically as type 3 deformities subsequently developed a proximal tibia epiphysis and thus did not represent true type 3 deformities. Fourteen limbs (11%) were characterized by global tibial deficiency but with proximal and distal epiphyses and could not be classified according to the Jones classification. Seventy-five of the 95 patients (79%) had associated anomalies. Other lower extremity anomalies were most frequent; however, upper extremity, spine, and visceral anomalies were also noted.

Conclusions: True type 3 deformity as described by Jones was not seen in our patient population; all patients developed a proximal epiphysis. Therefore, this group may be better served by limb salvage than amputation. Fourteen (11%) limbs, characterized by global tibia shortening relative to the fibula of variable degree, could not be classified according to the Jones classification. We propose adding this group as a new group within the Jones classification, which we call type 5. Finally, in this patient population, the Brown procedure for type 1 tibial deficiency universally failed, confirming results of prior studies.

Level of Evidence: This is a level IV study, a retrospective review of 95 patients with tibial deficiency from a single institution.

Key words: tibial deficiency, tibial hemimelia, Jones classification, Brown procedure

Copyright © 2015 Wolters Kluwer Health, Inc. Unauthorized reproduction of this article is prohibited.
birth, type 2 has a proximal but no distal tibia, type 3 has a distal tibia with the proximal tibia absent, and type 4 is characterized by diastasis of the distal tibia and fibula. In 1989 Schoenecker et al.² published a larger, multicenter series of 57 patients (71 limbs) in which all were classified according to the Jones classification. Both studies identified type 3 (no proximal tibia, but distal present) as the least common type of tibial deficiency. Kalamchi and Dawe³ sought to modify the Jones classification in 1985; however, this has not been widely accepted. In 2008, Weber⁴ reviewed a series of 63 patients and found that 15% of patients had deformities which did not fit into the Jones classification and proposed a new, 7-part classification system with additional modifiers (Fig. 2); however, this system also has not been widely accepted. A PubMed search of English literature since publication of the Weber classification revealed only 1 reference.

We reviewed cases of tibial deficiency treated at our institution from 1975 to 2012, with the goals of classifying these cases by the Jones classification, evaluating for the presence of associated anomalies, and categorizing the surgical treatments provided to these patients.

METHODS
This was an IRB-approved retrospective study of all cases of tibial deficiency treated at our institution since 1975. Patient charts and radiographs were examined to categorize patients by radiographic type (Jones classification), identify any recorded associated anomalies, and determine the patients’ full course of surgical treatment. Patients were excluded if they were treated before 1975, had incomplete records, or unknown full treatment course.

In our initial search of medical records, 15 had inadequate records or unknown final treatment. This left a study population of 95 patients, 30 with bilateral tibial deficiency, for a total of 125 affected limbs.

RESULTS
Morphologic Classification
Seventy-three of the 125 limbs (58%) were classified as Jones type 1A, 6 (5%) as type 1B, 18 as (14%) as type 2, and 12 limbs (10%) as type 4. Two patients had infantile radiographs suggesting type 3 tibial deficiency, that is, absent proximal but present distal tibia. However, in both the cases, overtime an obvious proximal tibial epiphysis articulating with the distal femur and with a distinct physis developed. Thus, we identified no “true” type 3 tibial deficiency patients in this patient population.

In 14 limbs (11%), the nature of the tibial deficiency could not be characterized according to the Jones classification. These limbs did, however, have a similar morphologic and radiologic appearance (Fig. 3A, B). All had global tibial deficiency with relative proximal and distal fibular overgrowth. A broad spectrum of deformity severity was seen in this group, varying from subtle deformity requiring no treatment to deformity sufficiently severe that knee disarticulation was performed. Two other morphologic features were often seen in these

FIGURE 2. Weber classification, black areas are bone, grey are cartilage. Reproduced with permission from Weber.⁴ Available at: http://link.springer.com/journal/11832/2/3/page/1 under a Creative Commons Attribution 2.0. Full terms at http://creativecommons.org/licenses/by/2.0.
patients: stable or resolving anterolateral bow (2 patients)
and/or great toe duplication (7 patients). Given the sim-
ilar morphologic appearance, we propose grouping these
cases into an additional subtype, which we refer to as
“Jones type 5.” The distribution of all patients by the
modified Jones classification is summarized in Table 1.

There were 30 cases of bilateral tibial deficiency in
our patient population. In 21 of 30, the Jones type was the
same on both sides, including 20 type 1A and 1 type 4.
Forty-nine of the 60 limbs in this bilateral group were
Jones type 1A and 29 of the 30 patients with bilateral
tibial deficiency had at least 1 limb classified at type 1A.
Nine patients had differing Jones types on either side. All
had 1 type 1A limb. The other limb was type 1B in 3
patients, type 2 in 3 patients, and type 5 in 3 patients.
These results are also displayed in Table 2.

Three of 95 patients had a first-degree relative with
tibial deficiency. These 3 patients all had type 1A defi-
cency and 2 of 3 had bilateral tibial deficiency. There
were also 8 of 95 patients who were adopted and therefore
have unknown family history.

**Treatment by Morphologic Type**
We reviewed treatment provided for all patients.
Sixty-two of the 73 type 1A limbs were eventually treated
with knee disarticulation. Of these, 47 were primary knee
disarticulations and 15 were performed after another
procedure failed, most commonly a Brown procedure
(fibular centralization and quadriceps reconstruction,
usually combined with a Syme amputation). Three pa-
tients underwent knee disarticulation subsequent to a
Syme amputation. Of the 11 patients who did not have
knee disarticulation, 2 had Syme amputation and knee

![Image](https://via.placeholder.com/150)

**FIGURE 3.** A and B, Proposed Jones Type 5 congenital tibial deficiency (see text). Both examples are characterized by global tibial
deficiency, proximal and distal tibial epiphyses, and relative proximal and distal fibular overgrowth. A, Anteroposterior radiograph
of a patient with a mild global tibial deficiency. B, Anteroposterior radiograph of a patient with a more severe global tibial
deficiency. Note medial ray duplication.

**TABLE 1.** Patient Distribution by Modified Jones Classification

| Type 1A | Type 1B | Type 2 | Type 3* | Type 4 | Type 5 |
|---------|---------|--------|---------|--------|--------|
| Patients | 53 | 6 | 18 | 2 | 11 | 14 |
| Limbs | 73 | 6 | 18 | 2 | 12 | 14 |

*We had no “true” type 3 patients in this series, as defined by Jones (absent proximal tibial epiphysis); both patients ultimately demonstrated the presence of a
proximal tibial epiphysis (see Fig. 3).

**TABLE 2.** Bilateral Patients by Modified Jones Classification

| Limbs | Type 1A | Type 1B | Type 2 | Type 3 | Type 4 | Type 5 |
|-------|---------|---------|--------|--------|--------|--------|
| Total (60) | 49 | 3 | 3 | 0 | 2 | 3 |
fusion; 4 had Brown procedures considered failures (1 nonambulatory patient and 3 with knee flexion contracture and no useful knee motion but no additional surgery), and 5 had no surgery. Of the patients who have not had surgery, 4 have planned knee disarticulation in the future and 1 is a nonambulatory spina bifida patient who is being treated nonoperatively. The failure of the Brown procedure in these patients confirms previous reports.2,6

Type 1B and type 2 patients were most commonly treated with Syme amputation and tibiofibular synostosis (Fig. 4). Three of 6 type 1B limbs and 11 of 18 type 2 were treated in this manner. Of the remaining 3 type 1B limbs, 1 was treated with Syme amputation, 1 with knee disarticulation, and 1 has yet to have surgery. Of the remaining 7 type 2 limbs, 3 were treated with Syme amputation, 3 with knee disarticulation, and 1 with below knee amputation and fibular excision.

Of the 2 patients whose infant films suggested type 3 deformity but who subsequently demonstrated the presence of a proximal tibial epiphysis (Fig. 5A, B), 1 was treated with knee disarticulation and the other with Syme amputation and tibiofibular synostosis.

The 11 type 4 patients (12 limbs) underwent Syme amputation (8), whereas 4 underwent limb salvage procedures with preservation of their feet.

The patients whom we categorize as type 5 had a spectrum of deformity and consequently treatment, which ranged from no surgery to knee disarticulation. Five patients in this group did not require surgery, 1 had excision of great toe duplication, 1 had rotational osteotomy with reduction of knee dislocation and excision of duplicated great toe, 3 underwent limb lengthening and reconstruction with circular frames, 3 underwent Syme amputation, and 1 had knee disarticulation.

Associated Anomalies

We evaluated all records for documentation of associated anomalies. Seventy-five of 95 patients (78%) had associated anomalies. The subset of patients with bilateral tibial deficiency had essentially the same rate of associated anomalies, with 22 of 30 patients (73%) having associated anomalies. These were most often other lower extremity deformities; however, upper extremity deficiencies, spine, and visceral anomalies were seen. A summary of associated anomalies by Jones type is shown in Tables 3 and 4.

There were a range of lower extremity anomalies seen. Deficiency of the lateral ray(s) was the most common lower extremity anomaly, affecting 20 patients. Medial ray and/or great toe duplication was seen in 13 patients. Ten patients had hip dysplasia or dislocation, and 10 patients had congenital femoral deficiency. Clubfoot of the contralateral limb was seen in 8 patients, tarsal coalition in 5, absence or hypoplasia of the medial ray in 5, femoral duplication in 4, lateral ray duplication in 3, and syndactyly in 3. Two patients had femoral condylar hypoplasia or absence and 2 had ipsilateral anterolateral tibia bow. The following deformities were seen in 1 patient each: opposite side fibular deficiency, opposite side lower extremity contracture, knee dislocation, cleft foot, coxa vara, and absent foot.

The most common upper extremity associated anomaly was cleft hand, seen in 15 patients. In addition, 8 patients had radial deficiency, 3 had absent long fingers,
2 had ulnar deficiency, and 1 each had isolated thumb hypoplasia, transradial deficiency, absence of hand, and elbow pterygium with single ray hand. Of the spine deformities there were, 8 congenital scoliosis, 4 tethered cord, 2 caudal regression/sacral agenesis, 1 lipomeningocele, and 1 myelomeningocele with kyphoscoliosis.

There were a range of other organ anomalies seen as well. Twenty cardiac anomalies were seen with the most common being ventricular septal defect (6 patients). Patent ductus arteriosus was seen in 4 patients, Tetralogy of Fallot in 3, atrial septal defect in 2, and valve abnormalities in 2 patients. Total anomalous pulmonary venous return, pulmonary stenosis, and truncus arteriosus were found in 1 patient each. Fourteen patients had gastrointestinal anomalies. Tracheoesophageal fistula was the most common, affecting 5 patients. Imperforate anus was found in 4 and abdominal wall defect in 2. Colon malrotation, duodenal atresia, and esophageal atresia were found in 1 patient each. There were 7 genitourinary abnormalities seen: 2 patients had 1 kidney, 2 had a horseshoe kidney, 1 had stacked kidneys, 1 undescended testes, and 1 inguinal hernia. Other anomalies seen included: hydrocephalus in 3 patients, ear malformation in 1, single-eye blindness in 1, Sprengel deformity in 1, and lipomatous mass on back in 1 patient.

There were 13 great toe duplications, including 7 of the 14 type 5 limbs, suggesting an association between medial duplication abnormalities and type 5 tibial deficiency. In addition, 2 patients of the 95 in our study had anterolateral bowing of the tibia, both with type 5 deformity. Thus, medial toe duplication with anterolateral bowing may be the presenting deformity of this type of tibial deficiency. We found no upper extremity duplications in our study population. All upper extremity deformities were deficiencies with cleft hands being the most common. Of the 15 patients with cleft hands, 10 had type 1A tibial deficiency, 1 had type 2, and 3 had type 4.

### TABLE 3. Patients With and Without Associated Anomalies by Modified Jones Type

| Type | Number With Other Anomalies [n (%)] | Number Without Other Anomalies |
|------|-----------------------------------|-------------------------------|
| 1A   | 61 (84)                           | 12                            |
| 1B   | 2 (33)                            | 4                             |
| 2    | 14 (78)                           | 4                             |
| 3 (revisited) | 2 (100)                    | 0                             |
| 4    | 7 (58)                            | 5                             |
| 5    | 12 (86)                           | 2                             |

### TABLE 4. Nature of Associated Anomalies by Modified Jones Type

| Type (Total Limbs) | UE Deformity | LE Deformity | Visceral | Spine |
|--------------------|--------------|--------------|----------|-------|
| 1A (73)            | 23           | 44           | 23       | 12    |
| 1B (6)             | 1            | 1            | 1        | 1     |
| 2 (18)             | 6            | 11           | 5        | 4     |
| 3 (revisited) (2)  | 1            | 2            | 0        | 0     |
| 4 (12)             | 4            | 5            | 2        | 0     |
| 5 (14)             | 2            | 12           | 2        | 1     |
| Total (125)        | 37           | 75           | 33       | 18    |

LE Deformity indicates lower extremity anomaly (other than congenital tibial deficiency); UE Deformity, upper extremity congenital anomaly.
suggests that cleft hand deformity may be most closely associated with type 1 deformity. There were 5 patients in our series with VATER syndrome and 1 with CHARGE syndrome.

DISCUSSION

The Jones classification system was published in 1978, based on a review of 20 patients and 29 limbs. This study of 95 patients and 125 limbs is the largest series of tibial deformity to our knowledge. We noted several interesting observations in this group.

First, no patient encountered in the 37-year interval of this study population had a true type 3 tibial deficiency. The 2 patients who in infancy had radiographs appearing to be type 3 subsequently developed radiographic confirmation of a proximal tibial epiphysis articulating with the distal femur, complete with a proximal tibial physis. Thus, based on our institutional experience, we caution that infantile radiographs suggesting type 3 tibial deficiency may in fact have an unossified proximal tibia, reminiscent of type IB (proximal tibia unossified at birth) deficiencies. Such patients likely warrant preservation of their knee, with distal reconstruction of the limb as appropriate for the severity of the limb deformity.

Second, 11% of the limbs in this study were not classifiable using the Jones classification system. Their deformity was characterized by global tibial shortening, with both proximal and distal tibial epiphyses evident from birth and relative fibular overgrowth. The severity of tibial shortening within this group represented a spectrum from sufficiently subtle to escape early detection to severe enough that foot a blation was performed to manage the relative limb length inequality. This group was also notable for a relatively frequent associated duplicated great toe, which may serve as a signal for the presence of the tibial deficiency. This group too had patients with a stable or resolving anterolateral bow of the tibia. This deformity has been previously described in the literature, either as a separate entity or as part of more comprehensive classification schemes. Specifically, this type was included in the Weber classification encompassing types I and V. However, we suggest that adding this type to the Jones classification system as “type 5” allows for greatest simplicity and comprehensive adaptation of a well-recognized and utilized classification system.

Third, the vast majority of our 73 cases of type 1A tibial deficiency ultimately required knee disarticulation. Other reconstructive options, specifically the Brown procedure, were universally ineffective in this group. This finding is in keeping with prior studies.

Tibial deficiency has been previously described as occasionally associated with syndromes and frequently associated with other anomalies. More recently, the genetic etiology of some of these conditions has been explored and mutations associated with tibial deficiency identified. In our series only 3 of 95 patients had family history positive for tibial deficiency, however, we found associated anomalies present in 75 of 95 patients. Some of these associated anomalies appear to be associated with specific forms of tibial deficiency. For instance, 10 of 15 patients with cleft hand deformity had type 1A tibial deficiency. Also, 7 of 13 great toe duplications were in patients with type 5 tibial deficiency. These types of associated anomalies may be helpful in future research identifying possible genetic or developmental etiologies of this disorder. It is also interesting to note that we did not find any upper extremity duplication deformities, although the lower extremity anomalies were a mix of duplications and deficiencies.

There is much still to be learned about this rare disorder and many areas for future study. The current study provides the largest review to date and helpful information both in the classification and treatment of these patients as well as areas for further research into etiology and associated anomalies.

REFERENCES

1. 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.
2. Schoenecker PL, Capelli AM, Millar EA, et al. Congenital longitudinal deficiency of the tibia. J Bone Joint Surg Am. 1989;71-A:278–287.
3. Kalamchi A, Dawe RV. Congenital deficiencies of the tibia. J Bone Joint Surg Br. 1985;67:581–584.
4. Weber M. New classification and score for tibial hemimelia. J Child Orthop. 2008;2:169–175.
5. Brown FW. Construction of a knee joint in congenital total absence of the tibia (paraxial hemimelia tibia). J Bone Joint Surg Am. 1965;47:695–704.
6. Loder RT, Herring JA. Fibular transfer for congenital absence of the tibia: a reassessment. J Pediatr Orthop. 1987;7:8–13.
7. Devitt AT, O’Donnell R, Fogarty EE, et al. Tibial hemimelia of a different class. J Pediatr Orthop. 2000;20:616–622.
8. Beck JJ, Altok J. Congenital tibial dysplasia with lateral bowing and duplication of hallux: case presentations. J Pediatr Orthop B. 2013;22:213–218.
9. Clark MW. Autosomal dominant inheritance of tibial meromelia: report of a kindred. J Bone Joint Surg Am. 1975;57:262–264.
10. Krugler L, Adbo R, Schwartz A. Tibial deficiency: a genetic problem. J Assoc Child Prosthet Orthop Clin. 1985;20:41.
11. Cho T-I, Baek GH, Lee H-R, et al. Tibial hemimelia-polydactyly-five-fingered hand deformity associated with a 404 G > A mutation in a distinct sonic hedgehog cis-regulator (ZRS): a case report. J Pediatr Orthop B. 2013;22:219–221.