

Tibial Hemimelia and Femoral Bifurcation

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abstract

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Femoral bifurcation and tibial agenesis are rare anomalies and have been described in both the Gollop-Wolfgang complex and tibial agenesis-ectrodactyly syndrome. This article reports a case of Gollop-Wolfgang complex without hand ectrodactyly. Tibial agenesis-ectrodactyly syndrome and Gollop-Wolfgang complex are variants of tibial field defect, which includes distal femoral duplication, tibial aplasia, oligo-ectrodactylous toe defects, and preaxial polydactyly, occasionally associated with hand ectrodactyly.

This article describes the case of a patient with bilateral tibial hemimelia and left femoral bifurcation. The proximal tibial anlage had not been identified in the patient's left leg. After failed fibular transfer procedure, the knee was disarticulated. The other leg was treated with tibiofibular synostosis and centralization of fibula to os calcis. At 7-year follow-up, the patient ambulates with an above-knee prosthesis and uses an orthopedic boot for ankle stability.

In patients with a congenital absence of the tibia, accurate diagnosis is of the utmost importance in planning future treatment. In the absence of proximal tibial anlage, especially in patients with femoral bifurcation, the knee should be disarticulated. Tibiofibular synostosis is a good choice in the presence of a proximal tibial anlage and good quadriceps function.



Figure: AP radiograph of a 10-month-old girl with left femoral bifurcation at the middle of the shaft and bilateral tibial hemimelia. There is no distal femoral epiphysis present on the left side.

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Tibial hemimelia is a rare anomaly causing marked leg shortening and severe equinovarus deformity of the foot.¹ The incidence of congenital deficiency of the tibia in the United States is approximately 1 per million live births.² Associated anomalies are common, such as skeletal abnormalities of the foot, polydactyly, femoral bifurcation, cleft hand, urogenital abnormalities, and radial hemimelia.³

Agensis of the tibia with bifurcation of the distal femur is an extremely rare anomaly. Such patients can present with abnormalities in 1 or both legs, with or without upper limb involvement.⁴ Distal femoral bifurcation has been described in familial cases of tibial agensis-ectrodactyly, and also as a part of Gollop-Wolfgang complex, which also includes tibial agensis, hand (foot) ectrodactyly, and femoral bifurcation.⁴

Jones et al¹ classified tibial hemimelia based on initial radiographs. Four types of deformity were recognized. In type I, the tibia is not visible. In type 1a, the tibia is completely absent with a hypoplastic distal femoral end. In type 1b, a rudimentary

tibia articulates with a relatively normal distal femoral end. In type II, the proximal tibia is preserved with a short tibial segment, but the distal tibial end is absent. In type III, the proximal tibia is absent. In type IV, a short tibia with distal tibiofibular diastasis is present.¹

The treatment of femoral bifurcation is simple—to resect at its base. However, the treatment of tibial hemimelia is challenging. Amputation, fibular transfer, or reconstruction procedures are the alternative treatment options.⁵ This article describes the results of our treatment and presents a case of Gollop-Wolfgang complex.

CASE REPORT

A 10-month-old girl presented with a grossly deformed left lower limb with a large antero-medial protuberance, just proximal to the knee. The fibula was palpable at the lateral aspect of both knee and ankle joints, but the tibia was not palpable at all. Both knees showed flexion deformity of 60°. Bilaterally, feet showed equinovarus deformity with all toes present. Radiographs demonstrated that the left femur

was divided in a Y-shaped fashion at the middle of the diaphysis. Total absence of tibia was noted bilaterally (Figure 1). During surgery, a proximal tibial anlage was found at the right knee along with the quadriceps tendon and patella. The right leg was classified as a type Ib longitudinal deficiency of the tibia, and the left leg was classified as type Ia.

At 1 year of age, the patient underwent fibular transfer procedures 3 months apart. Simultaneously, the bifurcated femur was resected. The right knee flexion contracture improved with a brace; however, the left knee remained with flexion deformity of 60°. Five months later, centralization of fibula to os calcis was performed at the right ankle. We suggested disarticulation of the left knee, but the family refused.

At 7 years of age, the patient was unable to walk. The left leg was disarticulated at the family's request.

At 8 years of age, the patient was ambulating with an above-knee prosthesis. Flexion deformity of the right knee was 20° (Figure 2). The right ankle was stiff with medial instability (10°), and she required the use of an orthopedic boot for ankle stability.

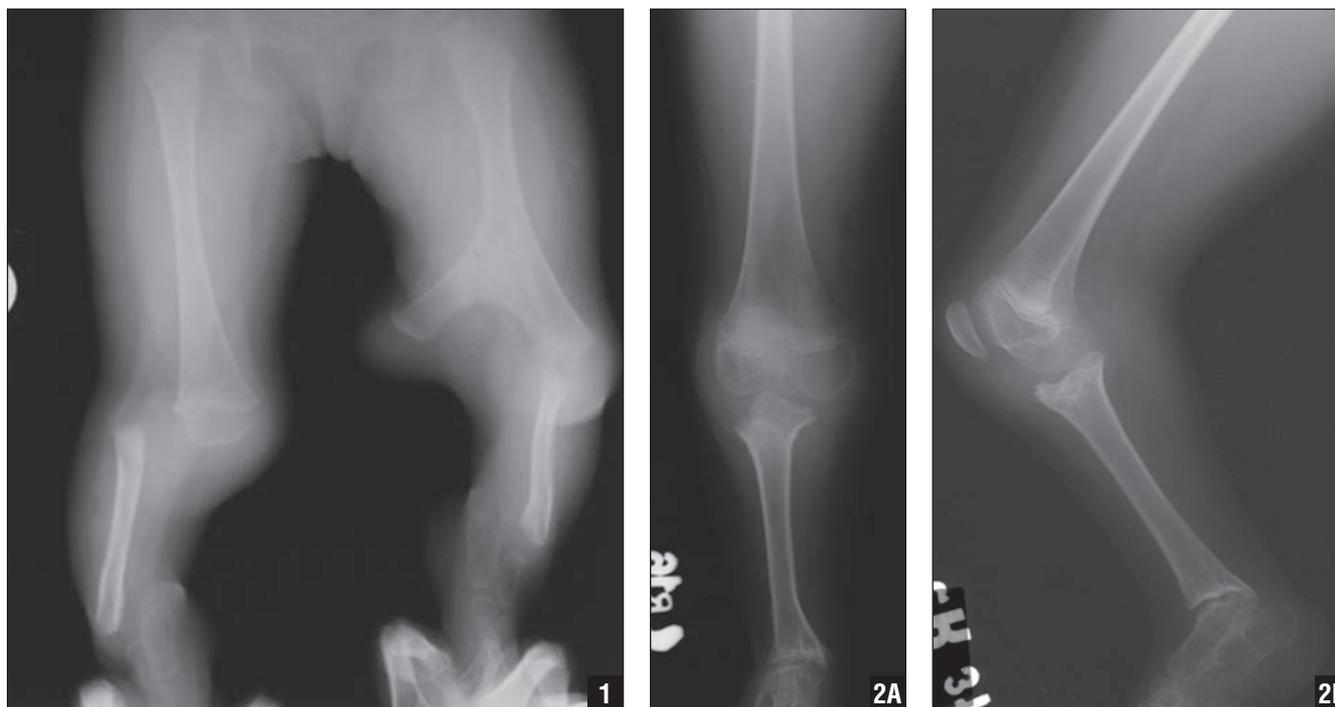


Figure 1: AP radiograph of a 10-month-old girl with left femoral bifurcation at the middle of the shaft and bilateral tibial hemimelia. There is no distal femoral epiphysis present on the left side. **Figure 2:** AP (A) and lateral (B) radiographs of the right lower limb at 7-year follow-up. The fibula has hypertrophied and its proximal end is flared to articulate with the femur.

DISCUSSION

Tibial agenesis-ectrodactyly syndrome and Gollop-Wolfgang complex are variants of tibial field defect, which includes distal femoral duplication, tibial aplasia, oligo-ectrodactylous toe defects, and pre-axial polydactyly, occasionally associated with hand ectrodactyly.^{4,6}

The other distinct type of limb developmental field is the fibular field, which is responsible for the development of the pubic portion of the pelvis, proximal femur, fibula, patella, anterior cruciate ligament, and lateral rays of the foot.⁶ The current patient appears quite similar to a patient with bilateral agenesis of the tibia and bifurcated femur, but without hand ectrodactyly, as reported by Bos and Taminiou.⁶ The term *Gollop-Wolfgang complex* is also used in patients with bifid femur and tibial agenesis without hand ectrodactyly.⁶

A cartilaginous tibial anlage has not been identified in the extremities of patients with femoral bifurcation; all patients have had a total absence of tibia without the presence of an extensor mechanism (type Ia).^{3,6-9} As was seen in our patient, knee flexion contracture has been reported to progressively increase in patients without a strong quadriceps.^{1-3,10,11} Thus, the treatment of femoral bifurcation with tibial hemimelia should involve the resection of

the anteromedial limb of the bifurcated femur and disarticulation of the knee at the end of the posterolateral limb.^{1,2,6,9-11}

If a cartilaginous tibial anlage is identified, any possible reconstruction of the lower leg should be considered.⁶ A type Ib deformity has good quadriceps function and should be treated with tibiofibular synostosis.^{2,10,11} The proximal fibula is embedded in the cartilaginous anlage of the tibia by the modified Brown procedure.^{2,10,11} Our case revealed some lateral knee instability but did not require an external support.

For the treatment of a type Ib deformity, the recommended distal procedure is Syme amputation.^{1,2,10,11} Centralization of fibula to os calcis does not resolve the problems such as leg-length inequality and ankle instability.¹¹ In our case, leg-length discrepancy did not occur because of the amputation of the other leg. However, the patient does need an orthopedic boot for ankle instability. ©

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