

Original Research

Received 20 June 2026

Revised 03 Aug 2026

Accepted 17 Aug 2026

Natural Resources for Human Health 2026, Vol. 6 (16s): 1389–1395

KEYWORDS:

Fibular hemimelia, FATCO syndrome, congenital limb deficiency, prosthetic rehabilitation.

Bilateral Fibular Hemimelia with FATCO Syndrome: A Rare Case of Complex Limb Deficiencies and Surgical Management

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ABSTRACT: Introduction: Fibular hemimelia (FH) is a rare congenital limb deficiency characterized by fibular aplasia or hypoplasia, often associated with tibial bowing, foot deformities, and digital anomalies. FATCO syndrome, a condition of unknown genetic etiology, is defined by fibular aplasia, tibial campomelia, and oligosyndactyly. Bilateral FH is even rarer and presents significant challenges in diagnosis, surgical management, and functional rehabilitation.

Case Presentation: We report a 5-year-old male with bilateral fibular hemimelia associated with FATCO syndrome. Clinical findings included absent fibulae, tibial bowing, oligosyndactyly, and severe foot deformities, with significant functional limitations in weight-bearing and mobility. Surgical intervention included Syme amputation on the right limb due to severe foot hypoplasia, and super ankle correction with tibial osteotomy and external fixation on the left limb to maximize residual function. Bilateral botulinum toxin injections were administered for muscle spasticity. Postoperatively, the patient demonstrated preserved functional mobility, requiring long-term prosthetic rehabilitation and physiotherapy.

Conclusion: This case highlights the complexity of bilateral fibular hemimelia and FATCO syndrome, emphasizing the need for early diagnosis, individualized surgical planning, and structured rehabilitation. While limb reconstruction may be considered in select cases, early amputation with prosthetic adaptation provides superior functional outcomes in severe deformities. Long-term orthopedic follow-up is essential to prevent secondary complications. Future research should focus on genetic factors, prosthetic advancements, and standardized treatment protocols to improve long-term outcomes in patients with complex congenital limb deficiencies.

INTRODUCTION

Fibular hemimelia (FH) is a rare congenital limb deficiency, with an estimated incidence ranging from 5.7 to 20 cases per million live births [1]. It is considered the most common congenital long bone aplasia, followed by defects of the tibia, ulna, radius, and femur in decreasing order of frequency [2]. The term "hemimelia" is derived from the Greek words *hemi* (half) and *melos* (limb), referring to the characteristic limb shortening associated with this condition [3]. The first documented case was described by Gollier in 1698, and later studies established that FH is not an isolated fibular deficiency but rather a complex disorder often associated with multiple musculoskeletal deformities [4].

FH can present unilaterally or bilaterally, with bilateral cases comprising approximately 30% of reported cases [2,5]. Additionally, it is frequently associated with other congenital anomalies affecting the ipsilateral limb, such as syndactyly, polydactyly, and radial dysplasia [2]. FH is most often sporadic, though it can occur as part of syndromic conditions [2].

Several classification systems have been proposed, with Achtermann and Kalamchi's classification being the most widely utilized. This system categorizes FH based on the degree of fibular deficiency [6]. In Type 1, the fibula is present but hypoplastic. Type 1A is defined by a proximal fibular epiphysis located below the tibial growth plate and a distal fibular epiphysis positioned above the talar dome. In Type 1B, there is a partial absence of the fibula, with no distal support for the ankle joint. Type 2 is characterized by a completely absent fibula, with severe tibial bowing that is more pronounced than in Type 1 cases.

The differential diagnosis of FH includes proximal focal femoral deficiency (PFFD), femurfibula-ulna syndrome, and femoral hypoplasia-unusual facies syndrome. Proximal focal femoral deficiency is characterized by a short, angulated femur with normal mineralization and may coexist with FH [7]. Accurate diagnosis is crucial for treatment planning and prognosis assessment. This case report presents a rare case of bilateral fibular hemimelia associated with FATCO syndrome, a condition of unknown genetic etiology characterized by fibular aplasia, tibial campomelia, and oligosyndactyly.

CASE PRESENTATION

A 5-year-old male, weighing 15.7 kg, presented to our orthopedic clinic with congenital lower limb deficiencies consistent with Fibular Aplasia, Tibial Campomelia, and Oligosyndactyly (FATCO) syndrome. He was born at 36+6 weeks gestation via emergency cesarean section due to breech presentation, with a birth weight of 2.5 kg. Antenatal ultrasound had detected lower limb shortening, and his mother had a history of hypothyroidism during pregnancy. There was no family history of skeletal abnormalities, though the parents were first-degree cousins.

At birth, the patient exhibited bilateral lower limb deformities, including absent fibulae, bowed hypoplastic tibiae, and oligosyndactyly, characterized by three fused toes on the right foot and four digits on the left, with three fused toes (Fig. 1). Additionally, he had femoral bowing and bilateral hip dysplasia, with no associated facial anomalies, cleft palate, or internal organ malformations.



Fig. 1. Anteroposterior (AP) X-ray of Bilateral Fibular Hemimelia. Demonstrating absent fibulae, tibial bowing, and oligosyndactyly with severe right foot osseous deficiency and femoral bowing.

Orthopedic findings and functional assessment

Comprehensive clinical evaluation confirmed bilateral fibular hemimelia, with right-sided tibial shortening and extensive foot malformation. The right foot exhibited near-total absence of osseous structures, while the left foot was partially developed with functional articulation (Fig. 2). Despite the limb deficiencies, knee extension and quadriceps function were intact, and ankle dorsiflexion was preserved bilaterally, though limited in the right ankle.



Fig. 2. Lateral X-ray of Right Lower Limb. Showing significant tibial bowing, absent fibula, and severe foot malformation with limited skeletal structure.

Given the complex skeletal deformities, genetic investigations were initiated to explore potential underlying syndromic associations. The differential diagnosis included FATCO syndrome, a rare congenital disorder of unknown genetic origin, and Fuhrmann syndrome, associated with WNT7A gene mutations. Given the constellation of fibular aplasia, tibial bowing, and oligosyndactyly without major upper limb involvement, the findings align most closely with FATCO syndrome. Fuhrmann syndrome, although sharing overlapping features, is typically associated with hand anomalies, which are absent in this case. Further genetic confirmation through WNT7A sequencing was considered.

Parental genetic counseling was advised, and targeted genetic testing was requested. To provide a concise overview of the patient's condition, a summary of key clinical features, investigations, and management strategies is presented in Table 1.

Table 1: Summary of case presentation and management

Category	Details
Demographics	5-year-old male, 15.7 kg
Birth & Family history	- Born at 36+6 weeks (breech, emergency C-section), birth weight 2.5 kg - Mother had hypothyroidism, no family history of skeletal abnormalities (parents are first-degree cousins)
Clinical features	Bilateral fibular hemimelia, tibial campomelia, oligosyndactyly (three fused toes on the right, four digits with three fused toes on the left), femoral bowing, bilateral hip dysplasia

Functional assessment	Intact knee extension and quadriceps function, limited ankle dorsiflexion (more severe on right), high risk for falls
Investigations	Genetic testing (FATCO syndrome suspected, WNT7A mutation considered), parental counseling recommended
Surgical management	- Right: Syme amputation, tibial osteotomy, flexion contracture release. - Left: Super ankle correction, tibial osteotomy, external fixation with 130-mm full and foot rings.
Adjuvant treatment	Bilateral botulinum toxin injections (200 units) for muscle spasticity
Postoperative outcome	No complications (no DVT, no excessive bleeding), preserved ADLs, require long-term prosthetic rehabilitation and physiotherapy

Surgical and functional management

A multidisciplinary team devised a staged surgical intervention plan aimed at optimizing limb function and mobility. The right lower limb underwent a Syme amputation to improve prosthetic adaptability, combined with a corrective tibial osteotomy and evaluation for flexion contracture release intraoperatively. The left lower limb was treated with super ankle correction, utilizing tibial osteotomy and external fixation. Additionally, bilateral botulinum toxin injections (200 units) were administered to address muscle spasticity, and a ring fixation system was applied for structural support, involving 130-mm full and foot rings bilaterally.

The complexity of limb deformity necessitated a careful assessment of limb salvage potential versus amputation. The decision for right-sided Syme amputation was guided by the severe foot hypoplasia, which limited weight-bearing potential. Conversely, the left-sided super ankle correction aimed to maximize residual function and facilitate prosthetic adaptation. Despite the severity of skeletal deformities, the patient demonstrated preserved functional mobility and engaged in age-appropriate activities of daily living (ADLs). However, given the inherent biomechanical instability, he was classified as high-risk for falls, necessitating long-term prosthetic rehabilitation and physiotherapy. His nutritional status remained adequate, and no perioperative complications such as deep vein thrombosis (DVT) or excessive bleeding were observed.

Long-term prognosis and follow-up plan

Postoperative rehabilitation will involve progressive weight-bearing training and dynamic prosthetic fitting. The patient will require periodic limb length monitoring, with potential future interventions such as guided growth modulation. Long-term prognosis remains favorable with appropriate prosthetic integration, though ongoing orthopedic surveillance will be essential to address potential complications such as knee valgus deformity or scoliosis due to asymmetric loading.

DISCUSSION

Fibular hemimelia (FH) is a rare congenital lower limb deficiency that varies in severity, from mild fibular hypoplasia to complete fibular aplasia with significant limb deformities. It often coexists with tibial bowing, foot malformations, and digital anomalies, posing significant challenges for surgical and functional management. In this case, bilateral fibular hemimelia associated with FATCO syndrome represents an exceptionally rare presentation, further complicating treatment strategies.

The etiology of FH and FATCO syndrome remains unclear, but it is believed to be influenced by genetic factors and developmental disruptions during limb formation [8]. Mutations in WNT7A, a gene essential for limb patterning, have been

implicated in Fuhrmann syndrome, a condition that shares overlapping features with FATCO syndrome [9]. However, Fuhrmann syndrome typically involves upper limb anomalies, which were absent in this case, reinforcing the likelihood of FATCO syndrome as the primary diagnosis [9].

The role of vascular disruptions in the embryological development of FH has also been proposed, particularly in cases where prenatal imaging detects lower limb shortening without identifiable genetic mutations [10]. As shown in a previous case report of a fetus diagnosed prenatally, ultrasonographic findings at 18 weeks of gestation revealed fibular agenesis, tibial angulation, and femoral shortening, suggesting that early detection is crucial for parental counseling and surgical planning [7]. Similarly, another prenatal case report documented bilateral FH in a fetus, which was confirmed postnatally and managed with bilateral Syme amputations and osteotomies to facilitate prosthetic fitting [11]. These findings highlight the importance of prenatal imaging and genetic analysis in suspected cases of limb deficiencies.

FH presents a wide phenotypic spectrum, and bilateral involvement, as observed in this case, is rarer and often more disabling than unilateral cases. Previous reports have documented severe bilateral FH cases with associated hip dislocation, femoral head necrosis, knee contractures, and valgus deformities, which significantly impact mobility and functional outcomes [10]. In our patient, bilateral fibular aplasia, tibial bowing, and oligosyndactyly presented unique challenges in terms of limb length discrepancy, joint stability, and weight-bearing capacity.

The severity of foot deformities is a critical factor in surgical decision-making [5]. In a previous report on a 16-year-old girl with severe bilateral FH, the patient exhibited valgus deformities, flexion contractures, and anteromedial tibial bowing, which required staged deformity correction using external fixators and Taylor Spatial Frames [12]. However, the outcomes of limb reconstruction procedures remain highly variable, with some cases still developing residual deformities, knee subluxation, or ankle instability despite aggressive correction.

The management of FH depends on multiple factors, including tibial integrity, foot structure, and functional prognosis [5]. In cases where severe foot hypoplasia prevents weight-bearing, amputation followed by prosthetic rehabilitation may provide a more functional outcome than limb reconstruction [5,13]. This approach was taken in our patient, where a Syme amputation was performed on the right limb due to severe foot hypoplasia, while super ankle correction was attempted on the left limb to preserve residual function [14].

A similar surgical strategy was reported in an infant with fibular hemimelia, where early Syme amputation facilitated more effective prosthetic fitting compared to repeated osteotomies and lengthening procedures, which often result in persistent deformities [15]. Likewise, another case report described a 5-year-old girl with a rare congenital limb deficiency, where a combination of prosthetic rehabilitation and ankle-foot orthosis enabled the child to achieve functional ambulation despite severe skeletal abnormalities [4]. These findings suggest that early amputation, when appropriate, may offer a superior functional outcome compared to prolonged reconstructive procedures in severe cases.

The long-term functional prognosis of FH is closely linked to prosthetic adaptation, rehabilitation, and orthopedic surveillance [13]. Our patient required progressive weight-bearing training, dynamic prosthetic fitting, and physiotherapy to facilitate age-appropriate mobility. A previous case of a 14-year-old girl with incomplete FH demonstrated that postural discrepancies, foot deformities, and tarsal coalitions can significantly impact long-term gait stability, underscoring the need for continuous orthopedic monitoring and adaptive interventions [16].

Given that bilateral FH presents greater biomechanical challenges than unilateral cases, early intervention and multidisciplinary care are crucial to optimize functional outcomes. Studies have shown that asymmetric limb loading in FH patients can contribute to progressive knee valgus deformities, scoliosis, and joint degeneration, necessitating long-term orthopedic follow-up and possible future corrective procedures [12]. In this case, potential future interventions such as guided growth modulation may be required to manage residual limb discrepancies and prevent secondary musculoskeletal complications.

CONCLUSION

This case of bilateral fibular hemimelia associated with FATCO syndrome highlights the complexities of congenital limb deficiencies and their management. Given the severity of the deformities, a combination of Syme amputation, super ankle correction, and external fixation was employed to optimize function. While limb reconstruction remains an option, early

amputation with prosthetic rehabilitation often provides superior functional outcomes in severe cases. Longterm orthopedic surveillance is essential to address potential complications, including joint instability and limb-length discrepancies. Future research should focus on genetic investigations, advancements in prosthetic technology, and standardized treatment protocols to improve patient outcomes in complex congenital limb deficiencies.

Declarations

Ethical approval

Not applicable

Funding

Not applicable

Conflict of interest statement

The authors declare no conflicts of interest or financial ties that could have influenced the work reported in this article.

Acknowledgement

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