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Case report

Bilateral Congenital Absence of the Femur: A Case Report

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Abstract
Congenital femoral deficiency (CFD) is a rare congenital limb anomaly ranging from mild shortening to complete absence of the femur. Bilateral complete femoral agenesis is exceptionally rare. We report a 3-year-old male born to a 42-year-old mother with poorly controlled type 2 diabetes. At birth, the patient was diagnosed with bilateral complete femoral agenesis, accompanied by multiple anomalies including bilateral clubfoot, cleft palate, congenital heart disease, bilateral cryptorchidism, and inguinal hernia. Radiographs revealed complete femoral absence with tibiae articulating directly with hypoplastic acetabula. The patient underwent bilateral posteromedial release for clubfoot, but recurrence occurred due to difficulty maintaining standard orthotic positioning. The severity and constellation of anomalies suggest a developmental insult during early embryogenesis, likely influenced by maternal metabolic factors. Traditional classification systems do not encompass this presentation, complicating diagnosis and treatment planning. Reconstructive options were not feasible due to the complete absence of femoral shafts; instead, orthopedic care focused on soft-tissue release, early bracing, and planning for prosthetic fitting to improve function. This case emphasizes the importance of maternal health during pregnancy and illustrates the critical role of individualized orthopedic management, including early soft-tissue procedures, custom bracing, and eventual prosthetic adaptation, in optimizing function in patients with severe limb deficiencies. It is the first reported case from North Africa.
Keywords. Bilateral Femoral Agenesis, Proximal Femoral Focal Deficiency, Pediatric Orthopedics, Limb Malformation, Case Report.

Introduction

Congenital femoral deficiency (CFD), including proximal femoral focal deficiency (PFFD), is a rare spectrum of developmental anomalies affecting the femur, with an estimated incidence of 1–2 per 50,000 live births [1]. The condition varies widely, ranging from mild femoral hypoplasia to complete absence of the femur, and is often associated with abnormalities of the hip, knee, foot, and surrounding soft tissues [2]. Most reported cases involve unilateral involvement, while bilateral complete agenesis of the femur is exceptionally rare, with only a few cases documented worldwide [3].

The etiology is thought to arise from disruptions in limb bud development during early embryogenesis (4–8 weeks of gestation), possibly triggered by genetic, vascular, or teratogenic factors [4,5]. Due to the rarity and severity of bilateral femoral absence, existing classification systems such as Aitken's and Paley–Guardo—commonly used for PFFD—are not fully applicable, complicating diagnosis and treatment planning [6,7].

We present a rare case of complete bilateral congenital femoral agenesis in a 3-year-old male with multiple associated anomalies, including bilateral clubfoot and congenital heart disease. To our knowledge, this is the first reported case from North Africa, contributing valuable data to the limited global literature on this severe form of limb deficiency.

Case Report

A 3-year-old male child, born full-term to a 42-year-old gravida 10, para 2 mother, presented with bilateral absence of the femur, noted at birth. The mother had a history of eight previous miscarriages and was known to have poorly controlled type 2 diabetes mellitus during pregnancy, despite transitioning from metformin to insulin in the first trimester. Folic acid supplementation was initiated only after the second month of gestation. There was no maternal history of radiation exposure or infections during pregnancy, and no family history of congenital anomalies was reported.

The child was delivered by elective cesarean section at term. At birth, multiple congenital anomalies were noted, including bilateral lower limb shortening, bilateral clubfoot, cleft palate, bilateral undescended testes, and inguinal hernia. On physical examination, the thighs were severely shortened bilaterally, with absent palpable femoral shafts. The child had equinovarus deformity of the feet, requiring support for postural stability (Figure 1).

Radiographic evaluation confirmed a complete bilateral absence of the femora, with the tibiae articulating directly with hypoplastic acetabula. No ossified femoral structures were identified (Figure 2). The diagnosis of complete bilateral congenital femoral agenesis was established.

Following bilateral posteromedial release for clubfoot, a foot abduction cast was applied as part of the initial orthopedic management to maintain correction and reduce the risk of relapse (Figure 3). Due to the complete absence of the femora, reconstructive surgery was not feasible. Management was therefore directed toward

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maximizing functional outcomes through early prosthetic fitting, aimed at supporting sitting balance and facilitating eventual assisted standing and ambulation.

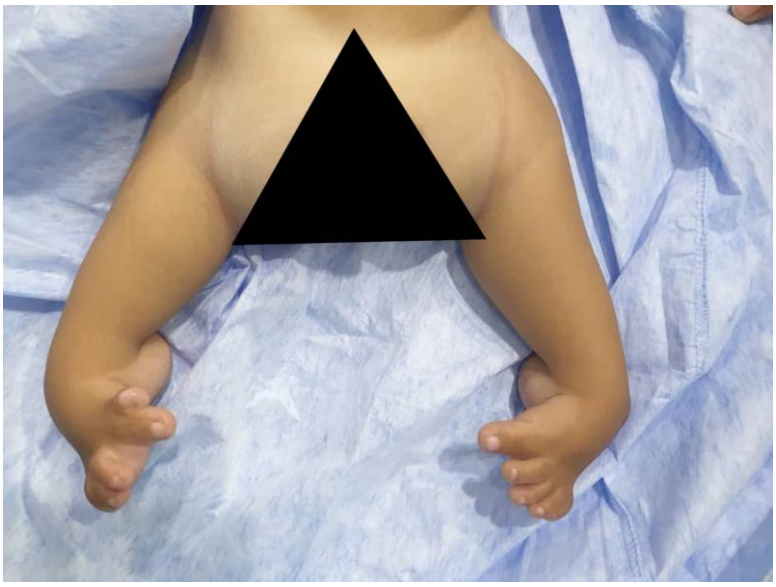

Figure 1. Clinical photograph showing severe bilateral lower limb shortening with equinovarus deformity. private areas digitally obscured.

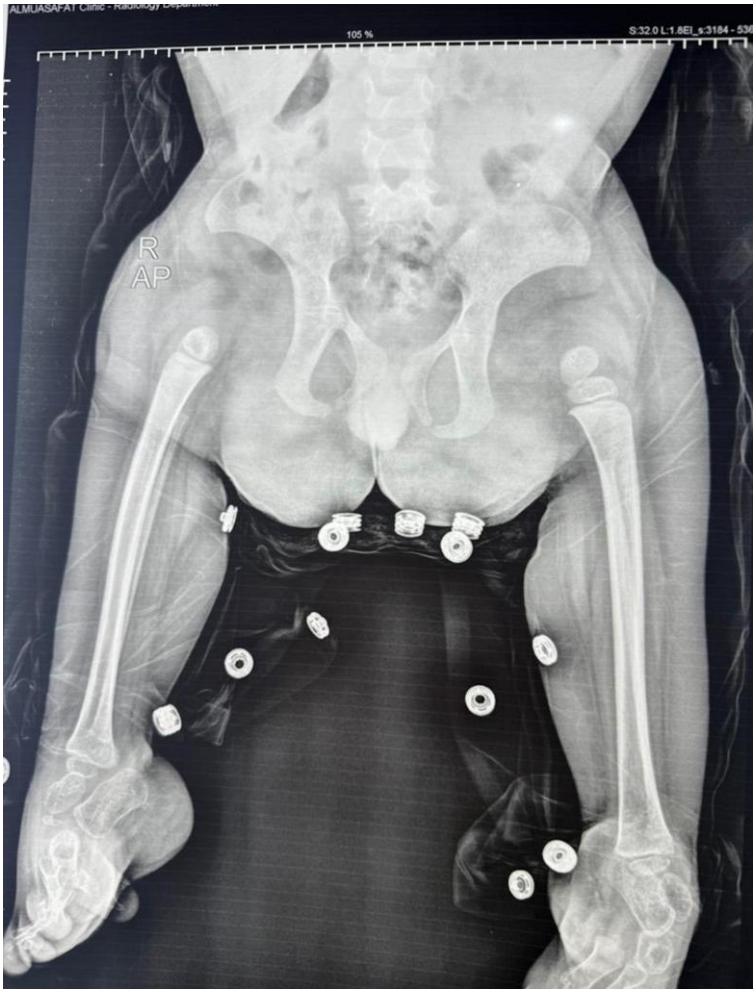

Figure 2 Anteroposterior radiograph of the pelvis and lower limbs showing complete absence of the femora with tibiae articulating with hypoplastic acetabula.

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Figure 3. Lower limbs immobilized in casts following bilateral posteromedial release for clubfoot.

A comprehensive physiotherapy program was initiated to enhance core stability and postural control. These interventions were essential to support independent mobility and developmentally appropriate functional progress. The patient is enrolled in a long-term follow-up program, with ongoing monitoring focused on mobility, growth, and functional adaptation. In the future, staged lengthening of the tibiae may be considered to improve prosthetic compatibility and enhance lower limb symmetry, depending on the patient's functional progress and skeletal maturity. At the time of reporting, the child can stand and walk with assistance, and can sit independently (Figure 4). Regular follow-up is ongoing to monitor growth, mobility, and overall developmental progress.

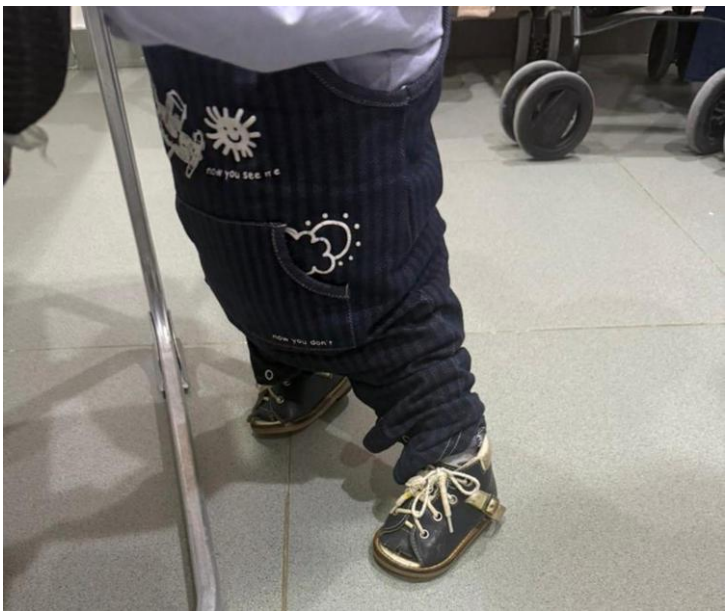

Figure 4 The patient is standing with assistance after cast removal.

Discussion

This case exemplifies an exceedingly rare presentation of bilateral congenital absence of the femur, representing one of the most severe forms of congenital femoral deficiency (CFD). CFD typically presents as a spectrum, from mild femoral shortening to complete absence. However, bilateral complete agenesis has been documented in only a handful of cases globally, with most reports focusing on unilateral or syndromic presentations [1,2].

This report represents, to our knowledge, the first documented case from North Africa, highlighting potential geographic underreporting. The patient, a 3-year-old male, was diagnosed neonatally with bilateral femoral agenesis and displayed multiple associated anomalies: bilateral clubfoot, cleft palate, congenital heart disease, bilateral cryptorchidism, and inguinal hernia. Such multisystem involvement aligns with literature reports of limb reduction defects accompanying systemic anomalies [3,4]. These findings suggest a global disturbance during early embryogenesis.

Limb development occurs between the 4th and 8th weeks of gestation, governed by critical signaling pathways such as FGF, SHH, and Wnt. Disruptions caused by genetic mutations, vascular compromise, or teratogens

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during this period can result in profound limb malformations. The co-occurrence of craniofacial, cardiac, and genitourinary anomalies supports an insult within this critical developmental window [5].

Radiographic imaging confirmed complete absence of both femora, with tibiae articulating directly with hypoplastic acetabula—an atypical anatomy not encompassed by current classification systems like Aitken or Paley–Guardo, which primarily address partial deficiencies [6,7].

Prenatal risk factors included poorly controlled maternal type 2 diabetes, advanced maternal age (42 years), and delayed folic acid supplementation. Maternal diabetes is a known teratogen linked with limb reduction defects and congenital heart anomalies, likely through oxidative stress and disrupted vasculogenesis during organogenesis [8–11].

Surgically, the patient underwent bilateral posteromedial release for clubfoot at age two, initially achieving correction. Recurrence occurred, possibly due to poor brace compliance—a known risk factor for relapse in congenital talipes equinovarus. The biomechanical instability caused by absent femora and knee joints further complicated the maintenance of correction [12].

Given the complete femoral absence, reconstructive surgical options are limited. Early prosthetic fitting remains the mainstay for improving seated balance, standing, and potential ambulation. A multidisciplinary approach—including orthopedics, cardiology, urology, rehabilitation, and developmental services—is vital to optimize function and psychosocial wellbeing [13]. This case underscores several critical points: The importance of preconception counseling and strict glycemic control in diabetic pregnancies, and the necessity of individualized, multidisciplinary care in complex congenital anomalies lacking formal guidelines.

Conclusion

This case presents a rare and severe manifestation of congenital femoral deficiency—complete bilateral absence of the femur—accompanied by multiple systemic anomalies. To our knowledge, this is the first reported case from North Africa, contributing valuable data to the limited global literature. The severity and rarity of this presentation challenge the applicability of existing classification systems and highlight the need for more inclusive frameworks to guide diagnosis and management. Given the lack of clear management protocols, especially for non-reconstructable cases, individualized care and early prosthetic planning are essential. This report also emphasizes the importance of maternal health and prenatal care—particularly glycemic control in diabetic pregnancies—to reduce the risk of congenital anomalies. The case underlines the necessity of multidisciplinary follow-up, customized bracing solutions, and long-term rehabilitation strategies to support functional outcomes and psychosocial well-being.

Patient Perspective

The patient's parents have acknowledged and accepted the congenital condition and expressed their satisfaction with the clarity of the medical explanations provided. They remain committed to long-term follow-up and multidisciplinary management to support their child’s development and well-being.

Informed Consent

Written informed consent was obtained from the patient’s legal guardians for publication of this case report and any accompanying images.

Ethical Approval

Ethical approval was not required for this case report, in accordance with institutional policies.

Author Contributions

Dr. Abdalsalam Andisha: Conception and design of the study, case data collection, clinical interpretation, and critical revision of the manuscript. Dr. Taimaa Langhi: Manuscript writing, figure preparation, and critical revision of the manuscript. Both authors: Conducted literature review, contributed to analysis and interpretation of findings, and approved the final manuscript.

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Conflicts of Interest

The authors declare no conflicts of interest related to this study.

Guarantor

Dr. Abdalsalam Andisha is the guarantor of this work and accepts full responsibility for the integrity of the case report and its contents.

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