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CASE REPORT

Prenatally Diagnosed Proximal Femoral Focal Deficiency: A Report of Two Cases

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Abstract: Proximal femoral focal deficiency is a developmental dysplastic congenital abnormality that primarily affects the proximal two-thirds of the femur, resulting in limb shortening and length discrepancies. The diagnosis is made by identifying a shortened femur with a proximal femoral deficiency. If left untreated, it can cause stunted growth, abnormal gait, spinal abnormalities, cosmetic concerns, behavioral changes, and psychological and emotional stress. We present two cases of women who sought routine antenatal care at our hospitals at 18 and 21 weeks of gestation. Both were diagnosed with isolated proximal femoral focal deficiency and chose to terminate the pregnancy, as the preferred management option, limb-lengthening, is not available in Ethiopia, and they did not want to pursue amputation and prosthetic therapy.

Keywords: proximal femoral focal deficiency, amputation, limb-lengthening, prosthetic therapy

Introduction

Proximal femoral focal deficiency is a very rare congenital condition, occurring in approximately 0.11 to 0.2 cases per 10,000 births, with only a limited number of case reports documented in the literature.[1] Proximal femoral focal deficiency (PFFD) is a developmental dysplastic condition that primarily affects the proximal two-thirds of the femur, resulting in limb shortening and length discrepancies.[2,3] The diagnosis is made by identifying a shortened femur with a proximal femoral deficiency. It can vary from mild limb shortening to the complete absence of the proximal femur and acetabulum.[3,4] Other possible features include abnormal limb rotation, incomplete iliofemoral joint formation, leg length discrepancy, and a varus deformity at the sub-trochanteric level.[3,5]

PFFD if untreated, it may result in stunted growth, abnormal gait, spinal abnormalities, cosmetic issues, and behavioral changes. In severe cases where limb-lengthening facilities are unavailable and amputation is required, the family may experience significant psychological and emotional stress. Therefore, prenatal diagnosis of PFFD is vital, as it helps parents make informed decisions about whether to continue the pregnancy, allowing them to explore all options and prepare emotionally.[6–8]

Ethical Review

Institutional approval was obtained both for the study of the case and for the publication of this case report, in accordance with institutional guidelines. Approval was granted by the Institutional Review Board (IRB) of Yekatit 12 Hospital Medical College (Y12HMC). Written informed consent for publication of this case report and any accompanying images was obtained from the mothers involved in the case.

Case Presentation

Case 1

A 31-year-old woman, G2P1, was referred to the Department of Obstetrics and Gynecology, Maternal Fetal Medicine unit at Addis Ababa University for a second-trimester anatomic scan at 18 weeks of gestational age. Her previous

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pregnancy was complicated by severe preeclampsia and intrauterine growth restriction at 29 weeks, leading to a stillbirth of a 750g fetus. The patient had no known chronic medical conditions and was otherwise healthy. For her current pregnancy, she had been taking 150 mg of aspirin daily, starting at 13 weeks GA. She lives in the city and has no history of drug exposure or radiation. The patient had no personal or family history of diabetes mellitus (DM), hypertension (HTN), or cardiac disease. The pregnancy was planned, wanted, and well-supported.

An ultrasound, performed using a Samsung HS40 device, revealed a shortened right femur measuring 11.6 mm, which was below the 1st percentile for gestational age according to the Fetal Medicine Foundation calculator, with no visible proximal epiphysis. The left femur measured 26.1 mm, falling within the 59th percentile for gestational age, and appeared normal. Limited limb movement was observed on the affected side, but the other long bones and craniofacial structures were normal (Figure 1).

The parents were informed of the findings and possible postnatal management options. After a thorough discussion, they decided to terminate the pregnancy. The outcome was a 230g male abortus with no significant craniofacial abnormalities (Figure 2). Consent for the procedure was obtained from the family, and a postmortem X-ray (Figure 3) confirmed the diagnosis of Proximal Femoral Focal Deficiency (PFFD).

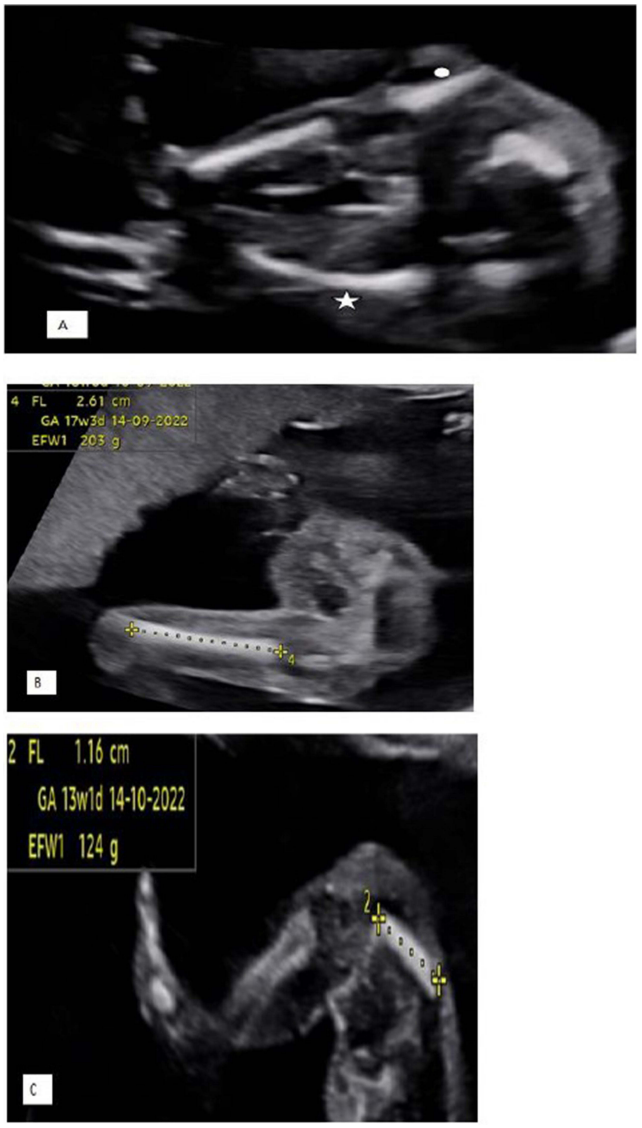

Figure 1 Ultrasound image (**A**) -shows both the normal femur (marked with a star) and the affected femur (marked with a circle) in the extended lower extremities. Image (**B**) -focuses on the normal femur, while image (**C**) -highlights the affected femur of the same fetus with PFFD.

Figure 2 Photograph of an abortus after expulsion, showing a significantly shortened right femur caused by PFFD.

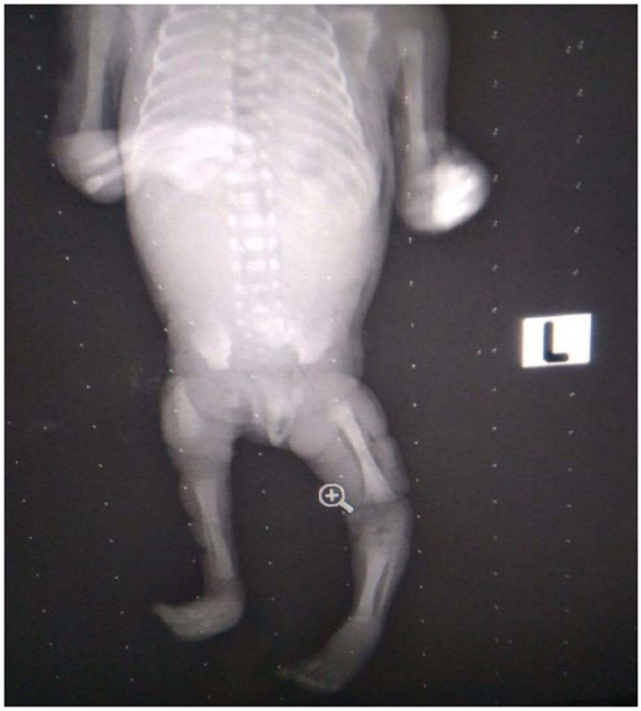

Figure 3 Post-expulsion X-ray of an abortus confirming a shortened right femur due to PFFD.

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Case II

A 25-year-old G2A1 woman, who did not recall her Last Normal Menstrual Period, her gestational age (GA) calculated from 13+1 weeks crown-rump length (CRL) ultrasound measurement. She was referred from a local health center to the Maternal-Fetal Medicine Unit at Abebech Gobena Referral Hospital at 22 weeks of gestation for a routine anatomical scan and continued care until delivery. All baseline investigations were normal. The patient had no personal or family history of diabetes mellitus (DM), hypertension (HTN), or cardiac disease. The pregnancy was planned, wanted, and well-supported.

An ultrasound performed using the Mindray DC-60 Exp device revealed a shortened left femur measuring 20.9 mm, which is below the 1st percentile for gestational age according to the Fetal Medicine Foundation calculator. The proximal epiphysis was not visible, but the lower leg and foot appeared normal. In contrast, the right femur measured 36.6 mm, within the 53rd percentile for gestational age, and appeared normal (Figure 4). All other long bones and craniofacial structures were normal (Figure 5–7).

The parents were informed of the results and potential postnatal management options. After a discussion, they chose to terminate the pregnancy. The outcome was a 330g male abortus showing a grossly shortened thigh with no significant craniofacial abnormalities (Figures 8 and 9). A postmortem X-ray was not performed due to the unavailability of X-ray services at the hospital at time of delivery, although consent for the procedure was obtained from the family.

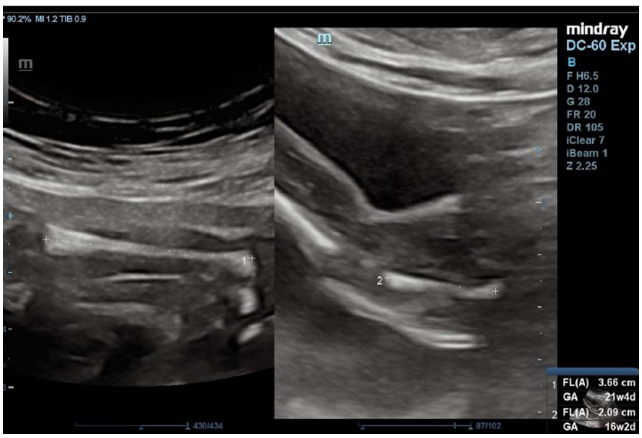

Figure 4 Dual-mode ultrasound of the femurs in a 22wk fetus complicated with PFFD. The left femur measures 20.9 mm, which is below the 1st percentile for gestational age, and the proximal epiphysis is not visualized. However, the right femur measures 36.6 mm, corresponding to the 53rd percentile for GA, and appears normal.

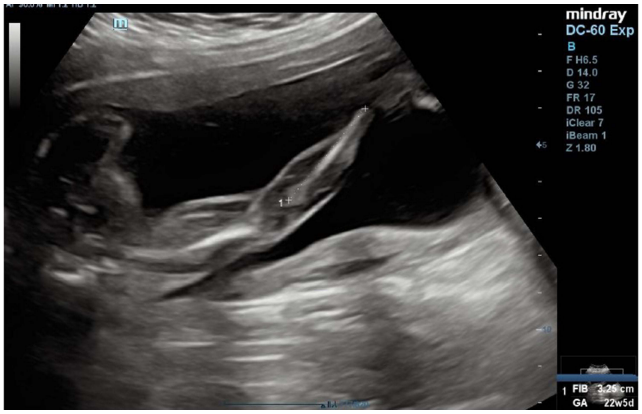

Figure 5 Three-segment lower extremity ultrasound of the same fetus, showing the right femur and fibula appear normal.

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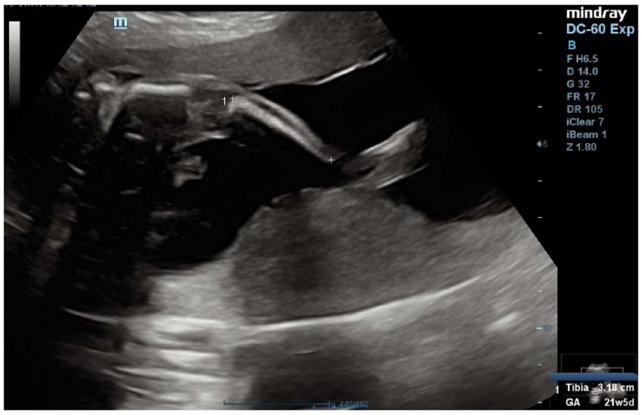

Figure 6 Three-segment lower extremity ultrasound of the same fetus, showing the left femur remains shortened, consistent with previous findings, while the tibia appear normal.

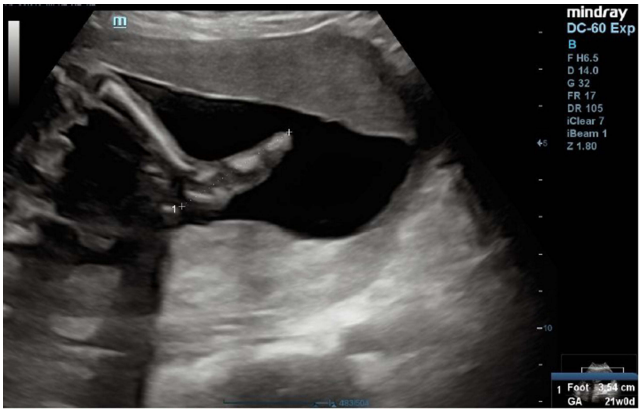

Figure 7 Ultrasound image of the tibia and foot of the left lower limb, showing normal development of both structures with no abnormalities, despite the shortened left femur.

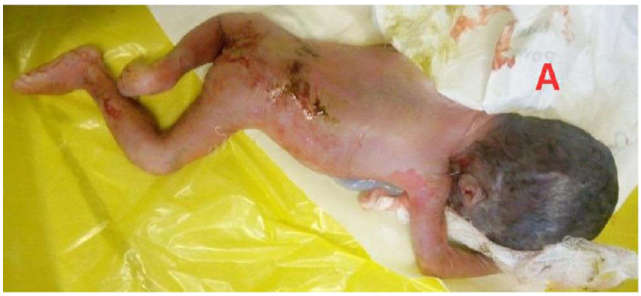

Figure 8 Postmortem images of the male abortus: (**A**) back view, showing a grossly shortened thigh with no significant craniofacial abnormalities.

Discussion

Proximal focal femoral deficiency (PFFD) is a rare congenital limb abnormality, occurring in approximately 1.1 to 2 cases per 100,000 live births. It primarily affects one limb in 85 to 90% of cases, though bilateral PFFD can also occur, with most diagnoses made after birth. Prenatal detection of femoral abnormalities is typically rare.[4,8–13]

PFFD is characterized by significant shortening, typically involving a partial skeletal defect in the proximal femur, often accompanied by a variable degree of hip joint instability. Associated anomalies may include congenital heart defects, clubfoot, spinal dysplasia, and facial dysmorphologies.[3,6,8] However, no associated anomalies were found in either of our cases.

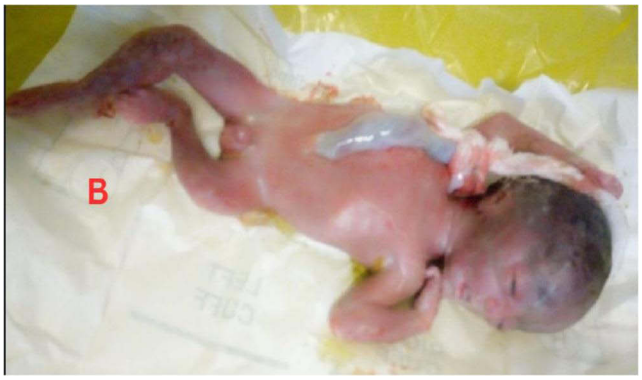

Figure 9 Postmortem images of the male abortus: (**B**) front view, showing a grossly shortened thigh with no significant craniofacial abnormalities.

When PFFD is suspected on ultrasound, it is important to rule out other congenital anomalies, such as femur–fibula–ulna syndrome, femoral–facial syndrome, and more severe conditions like thanatophoric dysplasia, achondroplasia, achondrogenesis, and chondroectodermal dysplasia.[1,14,15]

PFFD is classified based on the severity of the deformity. The most commonly used classification system, described by Aitken, consists of four categories (A–D), ranging from a mild form (A) to a severe form (D). This classification is based on factors such as the presence of the femoral head, hip joint stability, and the degree of acetabular hypoplasia (see Table 1 and Figure 10). [2,5,12,16–19]

Several factors have been suggested, including maternal use of thalidomide, viral infections, irradiation, trauma, focal ischemia, and diabetes. In our cases, the pregnant mothers did not have any of these known exposures.[1,8,20,21] While it is difficult to link PFFD to a specific gender, the right side is more commonly affected than the left, with an approximate ratio of 2:1. To avoid missing any skeletal dysplasia, it is important to measure both sides of the extremities according to ultrasound scan guidelines.[10,22] In our index cases, both fetuses were male, with the right femur affected in one case and the left femur affected in the other.

The management of prenatally diagnosed isolated PFFD is complex and influenced by factors such as associated abnormalities, available resources, planned postnatal care, and the family's social circumstances. Treatment involves a multidisciplinary and individualized approach, addressing aspects like leg-length discrepancy, proximal musculature condition, femoral rotation, and hip joint stability. Mild cases may be managed with limb salvage, lengthening, and hip reconstruction, while more severe cases could require amputation and prosthetic fitting. The majority of prenatally diagnosed cases result in pregnancy termination.[10,11,23–27]

Table 1 Aitken Classification System, Comprising Four Categories That Range from Mild (Type A) to Severe (Type D). The Classification Is Based on Factors Including the Presence of the Femoral Head, Hip Joint Stability, and the Degree of Acetabular Hypoplasia

Atiken Classification of PFFD				
Type	**Acetabulum**	**Femoral Head**	**Femoral Segment**	**Femoral Head-Shaft Attachment (Maturity)**
A	Present, normal	Normal	Short	Present and ossified; sub-trochanteric varus deformity
B	Mildly or moderately dysplastic	Present; delayed ossification	Short; proximal bone tuft, usually above the acetabulum	No osseous connection
C	Severely dysplastic	Absent or small; not ossified	Short; usually proximally tapered	No osseous connection
D	Absent: flat lateral pelvic wall	Absent	Short; deformed; often pointed proximally	Absent

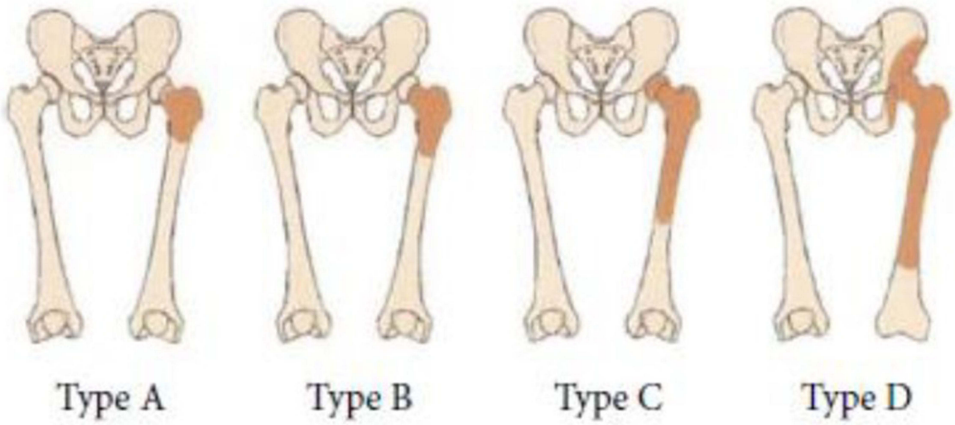

Figure 10 Aitken classification of PFFD, comprising four categories, ranging from mild (Type (A) to severe (Type D). The golden brown color represents the affected components of the femur.

Conclusion and Recommendation

PFFD is a rare congenital anomaly that can be diagnosed via prenatal ultrasound with a detailed assessment of both extremities. In resource-limited countries like Ethiopia, where limb-lengthening facilities are unavailable due to lack of the infrastructure, prenatal diagnosis is essential for formulating suitable management plans that can enhance prognosis and long-term outcomes for patients. The diagnosis of such fetal abnormalities may also prompt parents to consider pregnancy termination.

Data Sharing Statement

Upon reasonable request, the corresponding author will provide the data used to support the study's conclusions.

Acknowledgment

We would like to acknowledge and express our gratitude to all the management teams who were involved in overseeing the care of these mothers during their time at our hospital. Their dedication and teamwork played a vital role in ensuring the best possible care and support. Additionally, we would like to thank the mothers for their willingness to be presented as cases, their cooperation in providing the necessary data, and their commitment to attending regular follow-up appointments.

Author Contributions

All authors made substantial contributions to the reported work, whether that is in conception, execution, acquisition of data, analysis and interpretation, or in all these area; took part in drafting, revising, or critically reviewing the case report, provided their final approval for the version intended for publication, chose the journal for submission, and agreed to be accountable for all aspects of the work.

Disclosure

The authors declare that they have no conflicts of interest in this work.

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