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Case Report

A case report of proximal focal femoral deficiency diagnosed at birth in a term male neonate

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ABSTRACT

Proximal focal femoral deficiency (PFFD) is a rare congenital abnormality of the proximal femur and hip that can cause substantial limb-length discrepancy. Recognition at birth allows assessment of the affected limb and planning of long-term care. A male neonate born at 40 weeks of gestation by lower-segment caesarean section cried immediately after birth. His mother was unbooked and had not undergone antenatal ultrasonography. Examination revealed shortening of the left thigh, with the affected limb positioned in flexion, abduction and external rotation and an apparent limb-length discrepancy. Examination of other systems was within normal limits. Radiography showed marked deficiency of the left proximal femur with a high-riding residual femoral shaft, supporting a diagnosis of left PFFD in the immediate neonatal period. A shortened thigh on newborn examination should prompt radiographic evaluation. Further definition of proximal femoral anatomy and associated limb abnormalities is important for classification and treatment selection. Care planning should involve paediatric and orthopaedic teams, rehabilitation professionals and the family.

Keywords: Proximal focal femoral deficiency, Congenital femoral deficiency, Neonate, Limb-length discrepancy, Rehabilitation

INTRODUCTION

Proximal focal femoral deficiency (PFFD) is a congenital abnormality affecting development of the proximal femur and hip. Its manifestations range from femoral hypoplasia to extensive absence of the femur, with variable acetabular and joint abnormalities. The severity of the condition reflects both the skeletal deficiency and the functional potential of the affected limb.[1]

Recognition of an abnormally short thigh at birth provides an opportunity to define the anatomy before decisions about rehabilitation or reconstruction. Radiographs demonstrate the ossified skeleton, while magnetic resonance imaging (MRI) can characterize cartilage and soft tissues that are not adequately depicted on plain films.[2] We report left PFFD identified in a term male neonate and discuss its implications for assessment and multidisciplinary care planning.

CASE REPORT

A male neonate was delivered at 40 weeks of gestation by lower-segment caesarean section. The mother was unbooked, and no antenatal ultrasonography had been performed. The newborn cried immediately after birth. Shortening of the left lower limb, most evident in the thigh, was noted on examination at birth. The left lower limb was positioned in flexion, abduction and external rotation, with an apparent limb-length discrepancy (Figure 1). Examination of other systems was within normal limits.

A radiograph including the pelvis and proximal femora was obtained. It showed marked deficiency of the proximal left femur with a high-riding residual femoral diaphysis (Figure 2). Together, the clinical and radiographic findings supported a diagnosis of left PFFD in the immediate neonatal period. These findings establish proximal femoral deficiency but do not, by themselves, define the complete cartilaginous anatomy of the hip or the continuity between the proximal femur and residual shaft. Further anatomical assessment is important before assigning a definitive classification or selecting reconstructive treatment.[2]



Figure 1: Clinical photograph showing shortening of the left thigh and flexed positioning of the left lower limb. The photograph is limited to the lower body.

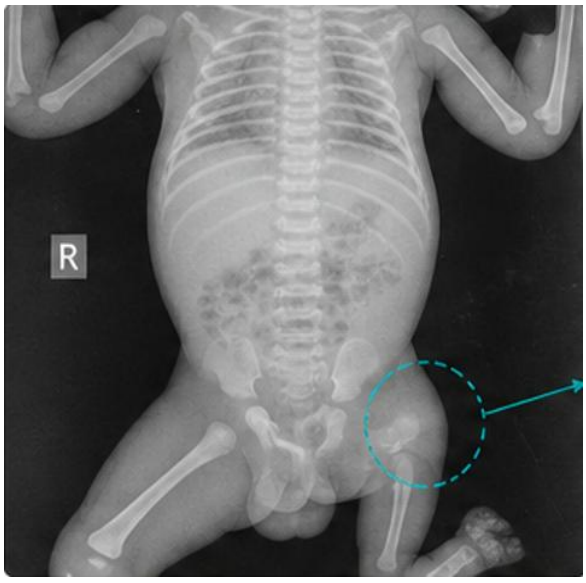


Figure 2: Radiograph showing marked deficiency of the proximal left femur with a high-riding residual femoral diaphysis. The right-side marker and the annotation identifying the left proximal femoral region are retained.

DISCUSSION

The shortened thigh and characteristic limb posture in this neonate provided the initial clinical clues to PFFD. Identification during newborn examination was particularly important because antenatal imaging had not been performed. Clinical recognition followed by radiography established the diagnosis at birth, allowing subsequent assessment to focus on anatomical severity and the functional potential of the limb. The pathogenesis of PFFD is not fully established. Histopathological study of an affected fetus demonstrated abnormalities in chondrocyte proliferation and maturation, supporting a disturbance of local skeletal development.[3] Etiological assessment should consider maternal illness, relevant exposures and family history without attributing causation in an individual child solely from the presence of the malformation.

The Aitken classification describes four groups according to the proximal femoral and acetabular anatomy.[4] In a neonate, absence of an ossified femoral head on a radiograph cannot be equated with absence of its cartilaginous precursor. MRI can help define the cartilaginous femoral epiphysis, acetabulum, head-to-shaft relationship and associated ligamentous abnormalities.[2] The radiographic appearance in this case therefore supports the diagnosis, while definitive anatomical classification requires further assessment. Assessment should extend to the entire affected limb. Associated abnormalities can involve the fibula, tibia, foot and stabilizing structures of the hip and knee, and may influence both reconstruction and prosthetic use.[1,2] A normal general systemic examination at birth does not establish that the deficiency is isolated. Evaluation for additional skeletal or systemic abnormalities should be guided by the clinical findings and any suspicion of a broader malformation syndrome.

Treatment is individualized according to femoral anatomy, hip and knee stability, foot function and the projected limb-length discrepancy. The paediatrician coordinates developmental surveillance and family counselling, while paediatric orthopaedic assessment defines reconstructive options. Physiotherapy and orthotic or prosthetic input support mobility as the child grows. Shoe lifts or orthotic support may be appropriate for selected children with smaller discrepancies.[1]

Children with suitable anatomy and stable or reconstructable joints may be considered for staged reconstruction and limb lengthening. External-fixator lengthening has produced useful gains in selected children with congenital femoral deficiency, but complications and the burden of repeated procedures require careful counselling and specialist follow-up.[5] In severe deficiency, prosthetic rehabilitation may provide a more appropriate functional strategy. Rotationplasty is an option for selected patients; long-term assessment of a small treated series found favourable overall function and

quality of life, although gait and postural differences persisted.[6] The management pathway should be discussed with the family over time as the child's anatomy, growth and functional needs become clearer. Early diagnosis supports this planning, but does not determine which intervention will ultimately be appropriate. This case illustrates the diagnostic stage of that pathway; assessment through childhood is needed to establish the eventual functional outcome.

CONCLUSION

PFFD should be considered when a newborn has a shortened thigh and a flexed, abducted and externally rotated lower limb. Radiography can establish proximal femoral deficiency at birth, while further assessment defines cartilaginous anatomy and associated abnormalities. Individualised care planning should integrate orthopaedic evaluation, rehabilitation and family counselling, with continued reassessment during growth.

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