

# Bilateral Proximal Femoral Focal Deficiency with fracture shaft of femur – A case report and review of literature.

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## Citation

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## Abstract

Proximal Femoral Focal Deficiency (PFFD) is a rare congenital deformity with femoral deficiency varying from hypoplasia with or without acetabular dysplasia to complete absence of the femur. It is bilateral in 15%. We report a case of congenital bilateral PFFD with fracture of shaft of femur.

## CASE PRESENTATION

A 13 weeks male child born in a twin pregnancy, born out of nonconsanguineous marriage was brought to us with gross deformity of lower limbs. The babies were born at full term by normal vaginal delivery. The labour had been difficult. The other sibling was born without apparent deformity but had died after three days. The baby in our case report was born with gross deformity of the lower limbs, the left leg being much shorter than the right leg. According to parents the baby was not moving the right leg at birth and cried on passive movements of right leg. They were told that the thigh bone on right side is fractured. None of the parents or their blood relatives had such a deformity

On presentation to us the left leg was much shorter than the right one. The right lower limb was kept in flexion and external rotation at the hip (Fig 1 & 2). The thigh on left side was markedly reduced in length with no solid structure felt on palpation. The baby was moving both the lower limbs. The feet were normal in shape and size. No other deformity was seen on examination of rest of the body. Radiograph of the baby (Fig 3 & 4) showed complete absence of femur on left side with united fracture of shaft of hypoplastic femur of right side with no ossific nucleus of head. There was no hypoplasia of fibulae.

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## DISCUSSION

Proximal focal femoral deficiency (PFFD) is a rare congenital anomaly characterized by failure of normal development of the femur (usually proximal femur) and hip joint. Significant variability in the clinical presentation and degree of deficiency is common<sup>1</sup>. Congenital abnormalities of the femur vary from a deficiency of the entire femur with abnormal development of the pelvis to a hypoplastic femur of normal configuration<sup>2</sup>. It may occur with or without fibular hemimelia (most common associated anomaly with PFFD seen in 2/3 of patients) and can be unilateral or bilateral (15%) in presentation. All muscles of thigh are present, but most are smaller than their normal counterparts (except obturator externus muscle)<sup>3</sup>. PFFD is commonly classified by Aitkin's classification which classifies PFFD as Class A (femoral head present with varus deformity), Class B (femoral head present with delayed ossification, varus and pseudoarthrosis may develop), Class C (absent femoral head with acetabular dysplasia and shortening of femur) and Class D (absent femoral head with severe dysplasia and severe shortening of femur). Kalamchi A et.al introduced a different classification based on initial radiographic appearance in 60 patients (70 affected limbs) in their series. They classified PFFD into 5 groups. Group I (short femur with good hip joint- 27%), Group II (short femur and coxa vara- 24%), Group III (short femur with proximal deficiency, a well-developed hip joint, and broad angulated and sclerosed diaphysis- 21%), Group IV (dysplastic distal femoral segment with no hip joint- 23%) and Group V (total absence of femur- 4%)<sup>4</sup>. Children with PFFD and their families are faced with many treatment decisions, both nonsurgical and

surgical. Nursing care is central in the care of these children and their families both for psychosocial support and teaching during the decision-making process and for being a patient advocate to help meet postoperative and rehabilitation goals<sup>5</sup>. Current management strategies aimed at improving functional ambulation are largely dependent on the degree of femoral shortening and the status of the hip and knee joint. Treatment of acetabular deficiency and proximal femoral deformity in cases of PFFD must be individualised. Reconstruction of the hip joint with pelvic and femoral osteotomies may be possible in mild cases of PFFD. Stability of the hip and knee joint must be achieved prior to consideration for limb lengthening strategies. Severe cases of PFFD may be beyond surgical correction and warrant

alternative strategies such as rotationplasty or selective amputation to facilitate prosthetic fitting<sup>1</sup>.

### **References**

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