Femoral Focal Deficiency: A Rare Case Report

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INTRODUCTION

Proximal focal femoral deficiency (PFFD) (congenital short femur) is a rare congenital anomaly occurs due to failure of normal development of the proximal femur resulting in limb shortening and pelvic abnormalities. The bony deformity presents as femoral deficiency and the focal lesion involve the proximal segment of the femur.¹ The etiology is unknown and no sexual predilection has been recognized.² There are several theories proposed and few postulated that the malformation is due to cellular nutritional disturbance at 4-6 weeks after ovulation where as others postulated a local vascular damage to mesenchymal tissue and while some mentioned intrauterine compression of the thigh at time of femoral diaphysis ossification. While no major role of heredity found.¹³ PFFD is usually unilateral, but bilateral cases are seen in 30% of affected individuals. It may be associated with other skeletal abnormalities like ipsilateral fibula Hemi Melia (most commonly associated), sacral agenesis, congenital absent patellar, proximal joint instability, lumbar and sacral spine deformities especially in infants of diabetic mothers. Associated skeletal abnormalities may be detected in approximately half the unilateral cases and 85% with bilateral involvement.¹²

Other anomalies may also be present in up to 70%, such as cleft palate, clubfoot, congenital heart defects, and spinal anomalies.⁴

CASE REPORT

A 3-month-old female child was brought to the out-patient department by her parents with complains of shortening of her left lower limb. General physical examination revealed shortening of left femur (Figure 1). No other abnormalities were present. Patient was born by normal vaginal delivery, and the pregnancy had been uneventful. Parents are healthy, not related and no family history of this condition or any other congenital anomaly was present. Parents of the child have one more child with no deformity. Mother has no history of miscarriages.

Laboratory tests were essentially within normal limits. Plain radiograph of both the lower limbs showed shortening of left femur and absence of proximal femoral component with the proximal aspect of shaft being v shaped indicating a pseudarthrosis (Figure 2). However, left side acetabulum was well formed.

DISCUSSION

PFFD is a rare congenital disorder resulting from the failure of the development of the subtrochanteric portion of the femoral shaft that is characterized by shortness, deformity, and dysfunction.³ Unilateral form is approximately 85-90% of all cases.⁶ Diagnosis is more often confirmed on radiological examination. A radiological classification

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proposed by Aitken is employed most widely both in diagnosis and therapeutic planning. Arthrography helps in establishing the degree of severity of the malformation, especially during the first 2 years of life. Magnetic resonance may demonstrate the entire disorder more effectively including any cartilaginous anlage and the status of the hip joint. The aim of management is to provide proximal stability, optimal function and cosmetic appearance. Standard orthopedic reconstructive procedures have proved totally ineffective in correcting leg length inequality seen in unilateral PFFD, especially when there is accompanying ipsilateral fibula Hemi Melia. In our case report, patient is classified according to Atiken as the proximal portion has tufted appearance with no osseous connection seen above it and falls into type C according to Aitken’s classification.

A radiological classification proposed by Aitken is employed most widely both in diagnosis and therapeutic planning. Gillespie and Torode identified two major groups for treatment purposes. Group I patients had a hypoplastic femur in which the hip and the knee were reconstructible and leg equalization was sometimes possible. Group II patients exhibited a “true” PFFD in which the hip joint was markedly abnormal. Although some of these patients had tenuous connections between the femoral head, and the proximal femur, the alignment and surrounding musculature were markedly abnormal. Furthermore, these legs were too shortened, rotated, and marred by flexion contractures of the hip and knee to be reconstructible. These patients required only reconstructive procedures that make prosthetic fitting easier.

Treatment is highly individualized and ranges from amputation and prosthetic rehabilitation to limb salvage, lengthening, and hip reconstruction.

Often no surgical reconstruction of any kind is indicated. Bilateral PFFD is best treated nonoperatively. These patients can walk without prostheses, but for social or cosmetic reasons extension prostheses may be provided. The patients learn to accept their short stature and are quite functional.

CONCLUSION

Proximal femoral deficiency is rare congenital deformity and treatment is individualized ranges from amputation and prosthetic rehabilitation to limb salvage, lengthening and hip reconstruction.

REFERENCES