Clinical Quiz

Submitted by: Yasir S. Siddiqui, MBBS, MS(Orth), Mazhar Abbas, MBBS, MS(Orth), Mohammad Zahid, MBBS, MS(Orth), Naiyer Asif, MBBS, MS(Orth).

From the Department of Orthopedic Surgery, Jawaharlal Nehru Medical College, Aligarh Muslim University, Aligarh, Uttar Pradesh, India.

Address correspondence to: Dr. Yasir S. Siddiqui, Department of Orthopedic Surgery, Jawaharlal Nehru Medical College, Aligarh Muslim University, PO Box 71, Aligarh 202002, Uttar Pradesh, India. Tel. +919837343400. Fax. +915712702758. E-mail: yassu98@gmail.com

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Congenital short femur

Clinical Presentation

A one-month-old male baby was brought to our outpatient department with the complaints of congenital shortening of his left thigh. The baby was first issue, born at term by spontaneous vaginal delivery. Antenatal history was typical. He cried immediately after birth. The parents denied any family history of skeletal abnormalities and consanguinity. At presentation, he was active, afebrile, with normal respiration, and his weight was 2.9 kg. Examination revealed shortening and deformaty of his left thigh. There was limitation of left hip and knee movements. There were no features suggestive of dysmorphism. The left thigh was placed in flexion and abduction. In addition, the ankle joint on left side was approximately at the level of the right knee (Figure 1). There was no other associated skeletal deformity, and rests of the systems were within normal limits. Laboratory investigations including complete blood count, erythrocyte sedimentation rate, serum calcium, serum phosphate and serum alkaline phosphatase were within normal limits. The antero-posterior radiograph of the left thigh with hip and knee was ordered as shown in Figure 2.

![Figure 1](image1.png) - Clinical photograph of patient showing shortened and deformed left thigh (arrow). Typical flexion-abduction contracture of hip with ankle joint almost at the level of contralateral knee.

![Figure 2](image2.png) - The antero-posterior radiograph of the thigh with hip and knee shows marked hypoplasia of proximal femur, bending of proximal femur at subtrochanteric region - subtrochanteric varus with pseudoarthrosis (arrow).
Clinical Quiz

Questions

1. What are the features evident on radiograph?

2. What is the diagnosis?

3. What are the associated skeletal deformities with proximal focal femoral deficiency (PFFD)?

Answers

1. The antero-posterior radiograph of the left thigh with hip and knee of the baby shows marked hypoplasia of proximal femur, bending of proximal femur at subtrochanteric region – subtrochanteric varus with pseudoarthrosis (arrow head).

2. Clinico-radiological diagnosis of patient is PFFD – left side. Shortened, deformed bulky thigh, typical flexion-abduction contracture of hip with ankle joint almost at the level of contralateral knee in association with radiological findings suggests diagnosis of PFFD.

3. Associated skeletal deformities with PFFD are flexion contractures of knee, hypoplastic patellae, absent or hypoplastic fibula, tibial shortening, tarsal coalition, equino-valgus deformity and absence of rays of foot.1,2

Discussion

Proximal focal femoral deficiency or dysgenesis of proximal femur or congenital short femur is a developmental disorder of the proximal femur and acetabulum ensuing in shortening of the affected extremity and impairment of the function.3 It is a continuum of inborn osseous anomalies characterized by a lack in the configuration of the proximal femur. The disorder is more frequently unilateral and is obvious at birth. The bony defect denotes the entity as a femoral deficiency, and this focal lesion always involves the proximal segment of the femur.1 The etiology is unknown and no familial or sexual predilection has been recognized.4 A radiological classification projected by Aitken is most widely used both in diagnosis and management.5 The PFFD is almost always an isolated occurrence. However, it may be associated with other skeletal abnormalities. Ipsilateral fibular hemimelia is the most commonly associated skeletal abnormality.4 The disorder can be readily diagnosed at birth due to its typical clinical presentation. Therapy is aimed at towards early and satisfactory ambulation and definite treatment depending on the severity.2 Remedial measures comprise fusion, osteotomy, lengthening procedures and amputation followed by the use of a prosthetic limb.

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References


