Proximal Femoral Focal Deficiency: A case report
Published on 12.02.2014

DOI: 10.1594/EURORAD/CASE.11400
ISSN: 1563-4086
Section: Musculoskeletal system
Area of Interest: Musculoskeletal bone
Procedure: Diagnostic procedure
Imaging Technique: Conventional radiography
Special Focus: Congenital Case Type: Clinical Cases
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Patient: 4 months, female

Clinical History:

A four-month-old female baby, delivered to non consanguineous parents, presented with right lower limb shortening since birth and was referred for radiographs of the lower extremities. There was no family history of similar affection.

Imaging Findings:

On anteroposterior radiograph of the lower limb, the right-side femoral head and proximal shaft appears deficient resulting in a short femur as compared to normal left-side femur. Mild flattening of the acetabular fossa is seen as compared to the left side. Also note, the absence of fibula on the same side and the deficient fifth metatarsal. The right tibia also appears relatively shorter in length as compared to the left and does not show ossification of proximal epiphysis which is seen on left side. Further screenings for various associated anomalies were carried out, and the baby was otherwise normal.

Discussion:

PFFD is rare and complex congenital anomaly. Aetiology of PFFD is not exactly known. Injury to neural crest cells that form precursors to peripheral sensory nerves L4/L5 or defect in proliferation and maturation of chondrocytes in proximal growth plate are two theories proposed [3]. Agents implicated in causing such injuries include anoxia, irradiation, ischaemia, infections, toxins and hormones. Thalidomide taken between the 4th and 6th week of gestation has been demonstrated to be definite cause [3]. The disorder is characterised by varying degrees of femoral hypoplasia resulting in limb shortening and pelvic abnormalities. The condition is bilateral in 15% of cases. The fibula may be absent in around 50-80% cases. The condition may be associated with other abnormalities like short tibia or fibula, patellar absence/hypoplasia/malposition, limb malrotation. There may be other associated anomalies like cleft palate, clubfoot, congenital heart defects and spinal abnormalities [1]. Hence, the patient needs to be screened for various associated abnormalities. There are several classification systems to grade the severity of femoral hypoplasia. The Aitken classification classifies into four classes in increasing order of severity as:

Class A: Shortened femur present proximally, ending at or slightly above the level of acetabulum. The femoral head is often absent or later ossifies. Femoral head presence is indicated by well-developed acetabulum. Additionally, there is a subtrochanteric defect which eventually ossifies and thereby establishes bony continuity. After ossification, there is usually subtrochanteric varus deformity.

Class B: A more severe defect or absence of proximal femur. This defect does not heal spontaneously. At skeletal maturity, there is no connection between the femoral head and proximal femur. The end of the proximal femur is above the acetabulum. The femoral head, although present, may have delayed ossification and there is often a bony
tuft on the proximal end of the shaft.

Class C: Absent femoral head does not ossify and a markedly dysplastic acetabulum. The femoral shaft is shorter than in a person with class B.

Class D: Severely shortened femoral shaft which often has only an irregularly ossified tuft of bone proximal to distal femoral epiphysis. No acetabulum is present with flat lateral pelvic wall. Our patient cannot be classified to any of this category as late ossification may occur. The aim of management is to provide proximal stability, optimal function and cosmetic appearance. The two important factors in management are lengthening of the affected femur at an appropriate age when the hip is stable and to provide pelvic-femoral stability [1, 2]. The various treatment options include a combination of different procedures like amputation, prosthesis, joint fusion, hip stabilisation and rotationplasty [1].

**Differential Diagnosis List:** Proximal Femoral Focal Deficiency, Complete shaft absence, Generalized dysplasia

**Final Diagnosis:** Proximal Femoral Focal Deficiency

**References:**


Description: AP radiograph of lower limbs, right femoral head, proximal shaft appears deficient. Mild flattening of acetabular fossa, absence of fibula seen with deficient fifth metatarsal. Tibia appears shorter, do not show ossification of proximal epiphysis. Origin: KIMS, Narketpally