Unilateral Isolated Proximal Femoral Focal Deficiency

A 3-day, full-term male infant with normal vaginal delivery was referred to us for lower limb discrepancy. Examination revealed that the right femur of the infant was considerably shorter than the left femur [Figure 1]. The distal epiphyseal region appeared normal. Measurements (lengths, structures, etc.) of all other long bones, head circumference and abdominal circumference were normal as per the age. No skeletal, thorax, cardiac, gastrointestinal, genitourinary and neurological abnormalities were observed. The facial profile of the infant was normal. The parents were healthy and nonconsanguineous. There was no family history of similar disease or any other disease. Moreover, there was no history of gestational diabetes, drug use, teratogen or radiation exposure, or a history of viral infection during the gestational period. X-ray examination confirmed the diagnosis of isolated, unilateral proximal femoral focal deficiency (PFFD) [Figure 2].

Isolated femoral hypoplasia (FH) is a rare congenital limb anomaly. It has an incidence of 1.1–2 in 100,000 live births. Mental disorders and chromosomal abnormalities are not usually present with PFFD. Surgical reconstruction results in a good prognosis. However, some cases of FH that include skeletal malformations may be accompanied by global dysplasia syndromes and termination of pregnancy may be the only reasonable option in such cases.[1] An important aspect is to determine whether isolated femoral abnormality is part of the syndrome.

Congenital hypoplasia of the femur has four uncommon malformations: PFFD, FH/unusual facial syndrome (FH/UFS), femur/fibula/ulnar hypoplasia and limb/pelvis-hypoplasia/aplasia syndrome.

It is caused by failure of the development of the subtrochanteric portion of the femoral shaft that is characterized by shortness, deformity and dysfunction. The most critical period for skeletal development is between the 4th and 8th weeks of gestation. The unilateral form occurs in approximately 85%–90% of all cases. It is usually sporadic, but few familial cases have been described. Its genetic transmission mode is unknown. Some etiological factors include poor diabetic control during the early weeks of pregnancy, drug exposure (thalidomide), viral infections, radiation, focal ischemia, chemical toxicity and trauma.

Various PFFD classifications have been made based on the relationship between the acetabulum and the proximal end of the femur. The most commonly used classification is the one described by Aitken and modified by Amstutz. Based on the presence of the femoral head, a stable hip joint or acetabular hypoplasia, it is classified in A–D. The Aitken classification does not consider the cartilage and soft tissue abnormalities.

FH/UFS is closely associated with diabetic embryopathy, bilateral FH and facial dimorphism (short nose, long philtrum, thin upper lip, small lower jaw and cleft palate).[2]

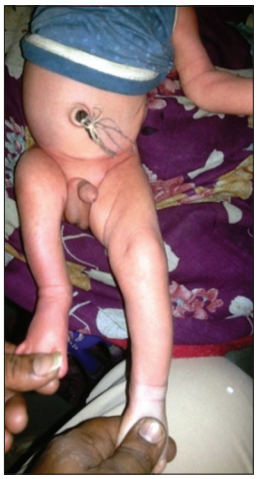

Figure 1: Clinical image showing lower limb discrepancy

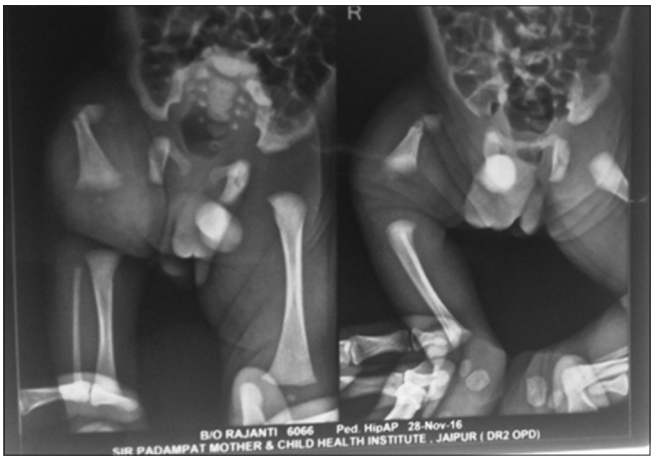

Figure 2: X-ray showing right proximal femoral focal deficiency

Femur, fibula and ulnar bone defects and various combinations of these were observed in the femur/fibula/ulnar complex. However, all extremities and many pelvic deformities are affected in autosomal recessive inherited limb/pelvis-hypoplasia/aplasia syndrome. In all, 30–60% of anomalies are associated with PFFD; these include fibular hemimelia and talipes equinovarus and rarely include oligodactyly, tibia bone bending, absence of the knee cross ligaments, spinal dysraphism and microcephaly.[3]

Although skeletal X-ray scans are useful for diagnosis, the condition may be accurately classified at the age of 1 or 2 years. Early neonatal ultrasound and magnetic resonance imaging are useful for the classification of PFFD.

In patients with simple FH, problem is limited to asymmetric legs. It does not usually develop a secondary deformity. If not corrected, PFFD results in an unpleasant appearance, excessive energy consumption during walking, scoliosis causing back pain and related symptoms. Surgical correction is required for significant shortness.

In PFFD type A, minimal side effects are observed, whereas types B, C and D require surgical correction. The aim of these surgical procedures is to synchronize the length of the leg, stabilize the feet and increase the pelvifemoral stability. After orthopedic correction, the long-term prognosis is usually good.[4]

PFFD is not associated with chromosomal abnormalities and patients have normal intelligence. Literature contains case reports in which termination of pregnancy has been selected before fetal viability.[2,5] The early recognition and exclusion of skeletal dysplasias should be done during the prenatal ultrasonography, and if required, treatment plans should be initiated and valuable information should be provided to the family.

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Conflicts of interest
There are no conflicts of interest.

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