Proximal Focal Femoral Deficiency in Classification of Proximal Focal Femoral Deficiency: Two Case Reports

Özet

Anahtar Kelimeler
Süt Çocuğu; Ultrasonografi; Kalça Eklemi; Femur; Alt Ekstremite Deformiteleri, Doğumsal

Abstract
Our purpose was to sonographically demonstrate the femoral heads of two infants with proximal focal femoral deficiency (PFFD) and to classify PFFD with the contribution of ultrasonography (US). On the radiograph of Case 1, a seven-month-old male with a short right thigh, there was marked hypoplasia of the proximal half of the right femur. Ipsilateral acetabulum was dysplastic. On the US, the femoral head was hypoplastic. On the radiograph of Case 2, a 16-day-old male with a short right thigh, the right femur was shorter and thinner than the left femur. An adequate ipsilateral acetabulum was found. US revealed a normal femoral head. US helped the classification of Case 1 as Aitken class C and Case 2 as and Aitken class A, by demonstrating the status of cartilaginous femoral heads in PFFD. Combined with radiography, US can be used in the classification of PFFD and in the evaluation of the unaffected hip.

Keywords
Infant; Ultrasonography; Hip; Femur; Lower Extremity Deformities, Congenital

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Corresponding Author: Ümit Yaşar Ayaz, Mersin Women's and Children's Hospital, Department of Radiology, Mersin, Turkey.
T.: +90 3242230701 GSM: +905377639442 F.: +90 3242230722 E-Mail: umityasarayaz@gmail.com
Introduction
Proximal focal femoral deficiency (PFFD) is a rare developmental disorder of the proximal segment of the femur and the acetabulum, resulting in a shortening of the affected limb and ensuing functional impairment. The incidence of PFFD was reported as one per 52,029 of the population (0.002%) [1]. According to the radiological classification proposed by Aitken [2], femoral head is present in class A PFFD, with an adequate acetabulum and a very short femoral portion. Initially, there is no bony fusion between that portion and the femoral head in Aitken class A PFFD. However, through skeletal growth and maturation, a bony fusion develops between the diaphysis of the femur and the head, neck, and trochanteric component. In Aitken Class C PFFD, the acetabulum is seriously dysplastic; the diaphysis of the femur is short with a tapered upper part; the femoral head is absent or very small with no connection with femoral diaphysis; and the femoral head (if present) never gets ossified [2]. Our aim in presenting the following two case reports was to ultrasonographically demonstrate the femoral heads of infants with PFFD, to present their plain radiography findings, and to classify them with the contribution of US data. Aitken's classification [2] was used as the reference.

Case Report 1
The male infant was seven months old on admittance, born at term after an uneventful pregnancy. No maternal risk factors including gestational diabetes or exposure to teratogenic agents during pregnancy were reported by the parents. Family history was unremarkable and there was no consanguinity. The physical examinations of the parents, including their limbs, were normal. The patient's parents were informed about the examination procedures and consent was obtained from them. All procedures were performed according to the World Medical Association Declaration of Helsinki (revised in 2000, Edinburgh). On physical examination of the infant, his right thigh was shorter than the left one and was bulky. The left leg, upper limbs, and face were normal. Physical examination, routine laboratory tests, and bone density evaluations revealed no systemic disease or rickets. On plain radiograph there was marked hypoplasia of the proximal 1/2 portion of the right femur with a tapered proximal end. The distal 1/2 portion of the right femur was thinner and shorter than that of the contralateral femur. The right acetabulum was dysplastic. There was a small notch superior to the right acetabulum, thought to represent a pseudoacetabulum formation. There was some superior displacement of the right femur; its tapered proximal end pointed to the pseudoacetabulum formation. On the left side, the acetabular angle was 41°, representing a relatively shallow acetabulum. Tibias and fibulas were present on both sides and no skeletal abnormality was observed in the left leg (Figure 1). On US images obtained by a 7.5 MHz linear-array transducer, a hypoplastic cartilaginous femoral head was demonstrated on the right side. Its diameter was 13.4 mm with no ossification center, while the left femoral head with an echogenic ossification center had a diameter of 19 mm. During the Barlow maneuver, US demonstrated that the right femoral head was superiorly displaced and located in an acetabulum-like depression on the iliac bone (Figure 2), concordant with the pseudoacetabulum detected on plain radiograph. The left femoral head coverage (FHC) was 50% in neutral position and decreased to 38% during the Barlow maneuver. The presence of a dysplastic acetabulum with an ipsilateral hypoplastic cartilaginous femoral head that was lacking an ossification center suggested that the PFFD in the present case had features consistent with Aitken class C.

Case Report 2
The male infant was 16 days old on initial admittance. He was born at term after an uneventful pregnancy. His nonconsanguineous parents reported no maternal risk factors including gestational diabetes or exposure to teratogenic agents during pregnancy. Family history was unremarkable. The physical ex-
aminations of the parents including their limbs were normal. The patient’s parents were informed about the examination procedures and gave their consent. All the procedures were performed according to the World Medical Association Declaration of Helsinki (revised in 2000, Edinburgh). On physical examination of the infant, his right thigh was shorter than the left one and was bulky. The left lower limb, upper limbs, and face were normal. His physical examination and routine laboratory tests revealed no systemic disease or rickets. On his first plain radiograph obtained on initial admittance, his right femur was shorter and thinner than the left femur. An adequate right acetabulum was found to be similar to the left acetabulum. There was a separate ossification center in the proximal portion of his right thigh representing the femoral neck (Figure 5), which was seen to have enlarged and to have fused with the more distal portion of the right femur four months later (Figure 4). Tibias and fibulas were present on both sides and no skeletal abnormality was observed on the left leg. On the last control radiograph obtained 12 months after the initial one, ossification centers of both femoral heads were visible and rather symmetrical. Right femoral length and thickness gradually increased and substantial improvement was observed. The right acetabular angle was 16° and the left acetabular angle was 30°, both within normal ranges (Figure 5). The first US examination performed by a 7.5 MHz linear-array transducer on initial admittance revealed a normal cartilaginous femoral head with no ossification center on the right side. Its diameter was 15.4 mm, close to the diameter of the left femoral head which was measured as 15.9 mm. During the Barlow maneuver, the right FHC was 22.6% (Figure 6), concordant with marked subluxation and the left FHC was 31%, higher than the right one but also concordant with subluxation. With the Ortolani maneuver both femoral heads returned...
to their resting positions. With the contribution of initial US findings, Case 2 was classified as Aitken class A. Four months later, control US revealed marked increase in FHC on both sides without any sign of overt subluxation, the right one being 48.2% (Figure 7) and the left one being 47.5% during the Barlow maneuver. On the last control US performed 12 months after the initial one, the right FHC was 49.2% (Figure 8) and the left FHC was 51% during the Barlow maneuver. Overt subluxation could not be detected on both sides. Eventually, follow-up data helped confirm the initial classification of the case as Aitken class A.

Discussion

The term PFFD is applied to a spectrum of conditions characterized by partial absence and shortening of the proximal femur(s). The pathology is mostly sporadic but familial PFFD cases have also been reported [3]. Aitken’s classification [2] is most widely employed in both diagnosis and therapeutic planning. Radiological features of PFFD are present at birth. Except in the most severe cases, the distal end of the femur is usually normal. In milder forms (classes A and B), the femoral head and neck ossify and fuse, but a non-progressive subtrochanteric varus deformity is constant. In more severe types, the acetabulum and the femoral head are absent, an iliac bony projection is observed lateral to and above the dysplastic acetabulum, and the femoral diaphysis is either rounded, pointed proximally, or almost completely absent. In all four forms, the femur is prominently shortened. After age two, evaluation of plain radiographs and appropriate assignment into one of the four Aitken’s classes become more feasible and more definitive than in infants below this age. The growth ratio of the abnormal to the normal limb throughout childhood is usually constant [4]. The Amstutz classification [5], another common classification system, has also been used in PFFD and has been applied to MRI [6].

To compensate for the inability of radiographs to demonstrate cartilage, MRI and US have been used to obtain more data in cases with PFFD [6–8]. Kayser et al. [7] reported that with US, the iliac line, femoral head, and greater trochanter could reliably be visualized in their patients with PFFD. In our cases, both the acetabuli and cartilaginous femoral heads could be evaluated using US, which helped us in classification of PFFD. Case 1 was a delayed case and the formation of a small pseudoacetabulum on the right side was thought to be due to a long-standing pressure effect of superiorly dislocated hypoplastic right femoral head on iliac bone. Though Case 1 had features consistent with Aitken class C, clinical follow-ups, control radiographs, and US examinations were recommended to the parents to verify the initial classification. Contact with the patient could not be established after the first examination, so we could not re-evaluate him. In Case 2, control US examinations revealed a normal-sized cartilaginous femoral head in its normal position on the affected side. We concluded that these features of Case 2 represented Aitken class A deformity, the mildest form of PFFD. Maldjian et al. [6] used MRI in diagnosis and classification of PFFD, stating that MRI was more accurate than radiographic evaluation for the classification of PFFD, particularly prior to the ossification of cartilaginous components in the femurs. MRI has perfect contrast resolution and it is superior to US in demonstrating all the bony elements (femoral...
head, femoral neck and shaft, acetabulum, iliac bone) and cartilage together, in clearly depicting their relationship with each other, and in showing any impingement [8], regardless of any limiting factor experienced with US, such as adverse effects of tissue thickness and acoustic shadowing caused by bone. However MRI is more costly than US and sedation is required in infants. Among all the imaging modalities, US seems to be the most practical method to evaluate non-ossified femoral head in PFFD. Like MRI, US is also free of ionizing radiation, but unlike MRI, it does not require sedation and it is relatively less costly. US also provides real-time images during maneuvers applied in dynamic hip examination. US is particularly useful as a rapid and practical tool in the very first diagnosis and classification of PFFD, prior to detailed MRI examinations which are stressful for the infant and the family. But experience in assessment of infant hips with US is essential; otherwise, misdiagnosis would be inevitable due to drawbacks in imaging. In our cases, combined with the radiographs, US was sufficient to evaluate the existence, size, and position of cartilage femoral head, both on the affected and unaffected sides. But we consider that MRI and US can be performed together, particularly in indeterminate cases, to combine the advantages of both techniques and to more confidently make the classification.

In conclusion, US was useful both in demonstration of cartilaginous femoral heads and in evaluation of their diameters/positions in our two infants with PFFD. Combined with plain radiography findings, US can be used in the classification of the affected femur and in evaluation of the unaffected side to rule out the coexistence of a possible developmental hip dysplasia.

Competing interests
The authors declare that they have no competing interests.

References

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