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Prenatal Detection of a Case of Proximal Femoral Focal Deficiency (PFFD)

Proximal femoral focal deficiency (PFFD) is a developmental disorder of the proximal segment of the femur and acetabulum, resulting in shortening of the affected limb and impairment of the function. It is a spectrum of congenital osseous anomalies characterized by a deficiency in the structure of the proximal femur.

It is a sporadic disorder with an increased incidence in infants of diabetic mothers. Abnormality ranges from aplasia/hypoplasia of the entire femur to complete absence of the proximal end. The disorder is more commonly unilateral and is apparent at birth; however, bilateral involvement is also rarely seen.

Keywords: Proximal Femoral Focal Deficiency (PFFD), Anomaly, Prenatal, Sonography

Introduction

Proximal femoral focal deficiency (dysgenesis of proximal femur or congenital short femur) is a rare congenital skeletal anomaly manifested by failure of normal development of a variable portion of the proximal femur.[1] The bony defect denotes the entity as a femoral deficiency, and this focal lesion always involves the proximal segment of the femur. In the past several years it was mainly detected almost always after birth by x-ray examination. Sonography is a noninvasive procedure for both the fetus and the mother and useful in the detection of fetal physical anomalies, of which PFFD is one of the rarest anomalies detected by this imaging method. The incidence of the deficiency ranges from 1 case per 50000-200000 population. Bilateral involvement is seen in 15% of the patients.

The etiology of this anomaly is unknown and no familial or sexual predilection has been recognized.[2] Various theories have been proposed.

Some of them postulated that the malformation is due to cellular nutritional disturbance at the time of cell division (four to six weeks after ovulation); whereas, others postulated a local vascular damage to the mesenchymal tissue and finally, others proposed the intrauterine compression of the thigh at the time of femoral diaphysis ossification. Heredity does not seem to play a major role.[1,3]

Case presentation

A 20-year-old primipara woman was referred to the imaging center for routine pregnancy ultrasonography with the use of high-resolution ultrasound unit (General Electric Logic α 500, Logic α 200) at 26th weeks of gestation.

Her marriage was not consanguineous, she had no infection or drug intake during early pregnancy and no congenital anomalies were detected in the past family history. At sonographic evaluation, a single 26-week male fetus with a short

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Received November 5, 2008;
Accepted after revision June 7, 2009.

Iran J Radiol 2009;6(4):253-256

right femur of 27mm in length was identified. The left femur length was 49mm (Fig. 1). The proximal portion of the right femur was partly absent and shortened, but the remaining part was normal.

The baby was delivered at 38-week gestation by caesarean section. Examination at birth revealed asymmetrical shortening of the lower limbs. There was no other dysmorphology. The rest of the skeleton was normal (Fig. 2A-C).

Discussion

PFFD is an uncommon but complex problem and a developmental defect of the proximal femur recognizable at prenatal survey, being the third most common longitudinal deficiency of lower extremity. PFFD may be detected prenatally with the ultrasound anomaly scan, which is usually performed after week 20. Abnormality ranges from aplasia/hypoplasia of the entire femur to complete absence of the proximal end. The etiology of PFFD is not exactly known, but certain theories have been proposed and agents implicated. Sclerotome subtraction is one such theory that has been proposed to explain several different limb deficiencies; specifically, this theory states that injury to the neural crest cells which form the precursors of the L4 and L5 peripheral sensory nerves, results in PFFD.

A second theory proposed by Boden et al. states that PFFD might be the consequence of an error in the proliferation and maturation of chondrocytes in the proximal growth plate. Such injuries could be caused by anoxia, ischemia, irradiation, bacterial and viral infections, toxins, hormones or may be mechanical, energy-induced or thermal injuries. Thalidomide when taken by mother from the fourth to the sixth week of gestation has been demonstrated to be a definite cause of PFFD in the human. Currently no evi-

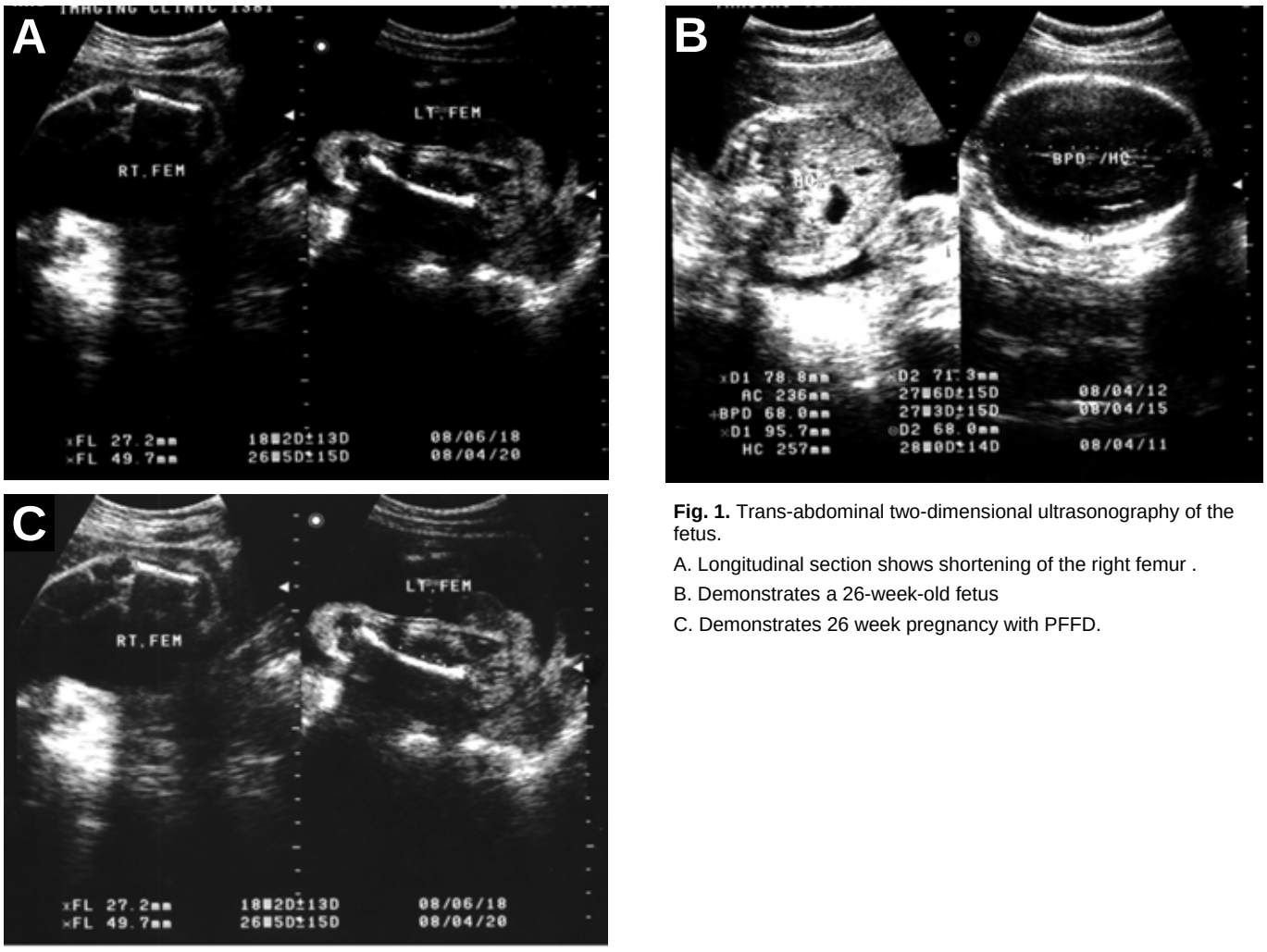

Fig. 1. Trans-abdominal two-dimensional ultrasonography of the fetus.

A. Longitudinal section shows shortening of the right femur .

B. Demonstrates a 26-week-old fetus

C. Demonstrates 26 week pregnancy with PFFD.

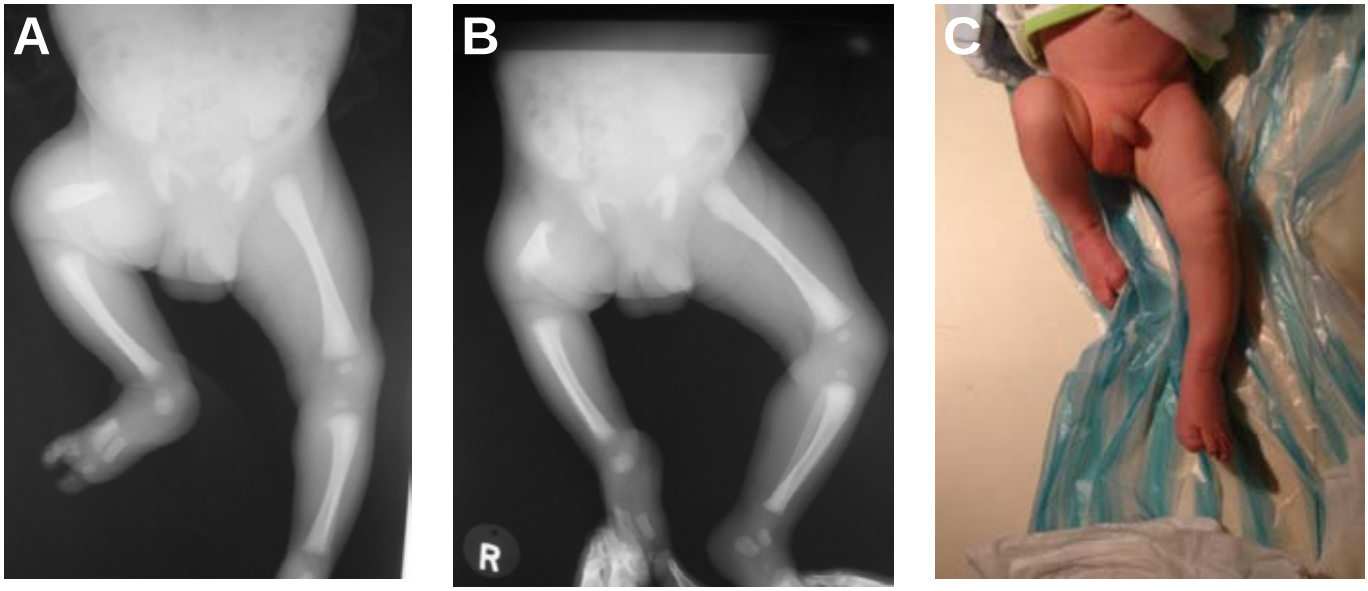

Fig. 2. A 26-week-old fetus with PFFD.
A&B. X-ray after birth showing short right femur.
C. Photographs of the child demonstrating a shortened thigh.

dence indicates a genetic etiology.

Aitken focuses on the status of the femoral head and neck:[4]

A: denotes the presence of a proximal femur that usually fuses to the shaft at skeletal maturity. Severe varus deformities may develop.

B: denotes a well-defined acetabulum but an unossified femoral head at birth. Severe varus deformities and pseudoarthrosis may develop.

C: denotes no femoral head and a poorly defined acetabulum and severe shortening of femur.

D: an extremely short or absent femur. Severe dysplasia of the acetabulum.

The appearance of PFFD is not subtle, so it is easily recognized. The femur is shortened, flexed, abducted, and externally rotated. Additionally, generalized knee hypoplasia has been reported.

Approximately 50% of patients with PFFD have other limb anomalies as well. Associated skeletal abnormalities may be detected in approximately half of the unilateral cases and 85% with bilateral involvement.[1,2]

A high incidence of fibular deficiency and valgus feet is associated with PFFD. Cleft palate, clubfoot, congenital heart defects and spinal anomalies occur rarely.

Marthese Ellul et al. studied a male newborn who was born by caesarean section. The parents were healthy and unrelated and there was no family history of this condition or any other congenital anomaly. At birth, he was noted to have a short right lower limb with an absent thigh. Plain radiography of both lower limbs showed an absent femur, fibula and acetabulum on the right side.[5]

Vishal Kalia et al. studied a four-year-old boy with gait disorder and shortening of the proximal part of both lower limbs since birth.

Plain x-ray of both lower limbs showed shortening of both femurs with thinning of both fibulas.[6]

Several surgical procedures are used to correct this anomaly. One limb-sparing procedure involves conversion of the knee to a lip joint by flexing it 90 degrees and fusing the femur to the pelvis. Another technique, developed by borggreve in 1930 called the "turn-about" procedure or "rotation-plasty" after an improvement by Van Hes, converts the foot into the knee joint; the limb is then fitted with a leg prosthesis.[7] Amputation is another option.

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