CASE REPORT

Bilateral Proximal Femoral Focal Deficiency in a Neonate: A Case Report

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SUMMARY

Proximal femoral focal deficiency is a rare and complex skeletal anomaly characterized by failure of normal development of the proximal femur, shortening of the lower limb, and pelvic abnormality especially the acetabulum. It was first described by Aitken in the late 1950s. It commonly occurs as a unilateral form but the bilateral variant is rare occurring in 10-15 % of cases. To the best of our knowledge, bilateral proximal femoral focal deficiency has not been reported in Nigeria. We, therefore, present this case in a 25-day-old neonate who presented to our hospital with features consistent with this complex skeletal anomaly.

Keywords: Bilateral; Proximal femoral focal deficiency; Plain radiograph; Nigeria.

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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[2]. However, distal femur is always present, thus, distinguishing it from femoral agenesis[1]. Since its description by Aitken[3], proximal femoral focal deficiency (PFFD) has attracted the attention of many clinicians[4].

The incidence of the PFFD ranges from 1 case per 50,000 to 1 case per 200,000 live births in the United States[5]. The incidence in Nigeria has not yet been reported to our knowledge.

Despite general theories, the presumed embryologic abnormality causing PFFD remains obscured. The aetiology is unknown and no familial or gender predilection has been recognized[6]. Some postulated that the malformation is due to cellular nutritional disturbance at the time of cell division (at four to six weeks' gestation). Others postulated a local vascular damage to mesenchymal tissue and some authors proposed intrauterine compression of the thigh at time of femoral diaphysis ossification[1, 7].

Proximal femoral focal deficiency is usually unilateral. However, bilateral disease has been

reported in 10-15% of individuals. The disorder is mostly an isolated occurrence but it may occasionally be associated with other skeletal abnormalities like ipsilateral fibular hemimelia (most commonly associated), caudal dysplasia, caudal regression syndrome, lumbosacral spine deformities especially in infants of diabetic mothers. Skeletal abnormalities may be detected in approximately half of the unilateral cases and 85% with bilateral involvement[1, 6]. Absence of the lateral foot rays have also been reported[1].

The diagnosis is often made by radiological evaluation which includes identification and description of PFFD and evaluation of associated limb anomalies by plain radiographs. Contrast arthrography or Magnetic Resonance Imaging is indicated when plain radiographic features are questionable and to disclose the presence and location of the femoral head[8].

This report presents a very rare case of bilateral proximal femoral focal deficiency in a 25-day-old female neonate.

Case Presentation

K.I.B. was a 25-day-old female neonate who was brought by her mother to the special care baby unit (SCBU) of the Paediatrics department, University of Maiduguri Teaching Hospital (UMTH) with complaint of shortness of both lower limbs which was noticed at birth. The patient was delivered to a 32-year-old para 7-mother (all alive and well). The mother did not know she was pregnant until 5 months gestation because she used to have lactational amenorrhoea in her previous nursing. Her last childbirth was two years ago. She did not attend antenatal clinic (ANC) in all her pregnancies due to ignorance of the importance of ANC. She admitted to have taken several traditional medications during the first trimester of this pregnancy because of occasional headache, fever and abdominal pains. She does not know any of these traditional medications by name. The index pregnancy was carried to term and delivered through spontaneous vaginal delivery at home with the help of a traditional birth attendant. The patient is the 8th child in a polygamous family setting. There was no family history of congenital anomaly. The parents denied any family history of diabetes or hypertension. The mother was a full-time housewife while the father was a farmer.

On examination the patient was afebrile, not pale, not jaundiced, acyanosed, no dysmorphic facie and was seen sucking well from the breast. Patient's weight was 3.7kg and had occipitofrontal circumference (OFC) of 35cm. Lower limbs examination revealed shortened, bulky thigh bilaterally (7cm on the right and 8cm on the left) with normal legs and feet. The total lengths of both lower limbs were 21cm and 23cm on the right and left respectively. However, the patient's father refused us the privilege of taking a still photograph. Other systems examinations were essentially normal. Laboratory tests including full blood count (FBC) and Electrolytes, Urea and Creatinine (EUC) were also within normal limits.

Plain radiographs of both lower limbs (Figures 1 and 2) showed shortening of the femur bilaterally which were displaced laterally with absence of femoral head on both sides. The proximal ends of the femora were tapered with flattened or dysplastic acetabuli. The remaining lower limb bones, joint spaces and soft tissues appeared normal. Based on the plain radiographic features a diagnosis of bilateral PFFD was made. The patient's parents insisted to go for traditional treatment and the case was lost to follow-up.

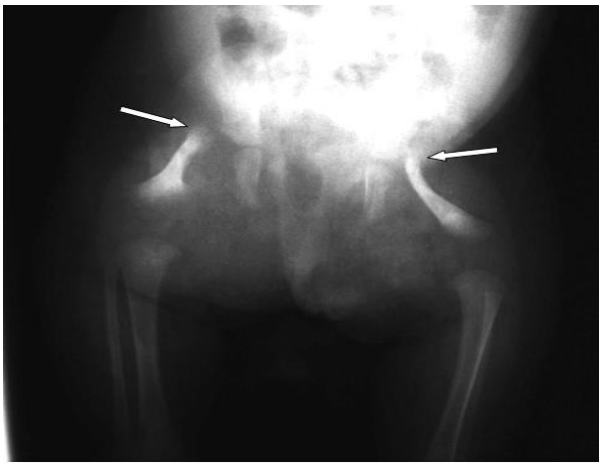

Figure 1: Plain AP radiograph of lower limbs showing shortening of the femur bilaterally which were displaced laterally (white arrows) with absence of femoral head on both sides. The proximal ends of the femora are tapered.

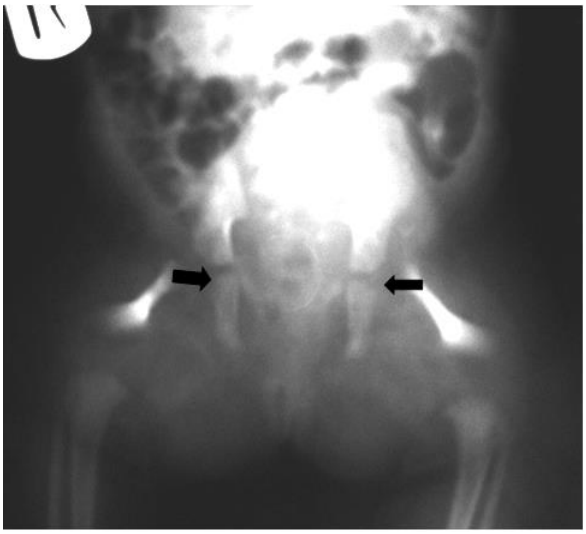

Figure 2: Plain AP radiograph of lower limbs. Note the flattened acetabuli, more severe on the left (black arrows) which signify dysplastic acetabuli.

Discussion

Congenital anomalies of the proximal femur with or without involvement of the hip joint have been observed and reported for many years [3]. PFFD is a rare form of congenital anomalies that affects the proximal femur and frequently the acetabulum[9]. It is also known as dysgenesis of the proximal femur and congenital short femur[10]. It is usually apparent at birth due to shortening of the lower limb as reported in this case. PFFD can be unilateral or bilateral[1, 6]. This case presented at birth with the bilateral form which is rarely seen compared to the unilateral form of PFFD.

Although there is an association between PFFD and other skeletal defects, most frequently ipsilateral fibular hemimelia, the precise aetiology of PFFD remains somewhat obscured[10]. Apart from caudal dysplasia or caudal regression syndrome, in which PFFD may be one of the congenital skeletal anomalies seen with lumbosacral spine deformities in infants of diabetic mothers, PFFD is almost always an isolated occurrence[6]. Thalidomide is known to cause PFFD when the mother is exposed to it in the fifth or sixth week of pregnancy, and it is speculated that exposure to other toxins during pregnancy may also be a cause. Other aetiologies that have been suggested, but not proven, include anoxia, ischaemia, radiation, toxins, bacterial and viral infections, chemicals, hormones, thermal and mechanical forces[2]. In the case presented the mother was said to be not a known diabetic or diabetic. However, there was history of the mother taking several traditional medications during the first trimester of this pregnancy. This may be implicated as the cause of the PFFD in this case. These traditional concoctions may have toxic or teratogenic effects on fetal development especially during the early phase of pregnancy[11].

The clinical features of PFFD are not subtle, so they are easily recognized. The femur is shortened, flexed, abducted, and externally rotated. Because of the short femur and bulbous thigh, examination of the hip can be difficult[3]. This case presented with shortened, bulky thigh bilaterally in addition to flexion at the hip and abduction of the thigh.

Although the clinical diagnosis of PFFD is usually straight forward, correct classification may be difficult solely on clinical findings[5], and therefore, is often made by radiological examination[8]. The diagnosis and classification of PFFD have been based mainly on plain radiographic findings and even though this method does not permit definite classification during the first year of life[5, 8]. The diagnosis of PFFD in this case presented was made using plain radiography.

Several classification schemes for PFFD based on anatomic relationships between the acetabulum and the proximal end of the femur have been proposed[8]. However, a radiological classification by Aitken[3] is probably the most often cited which proposed four classes (A-D) as follows:

Class A: Is the least severe type where the femoral head is present and attached to the shaft by the femoral neck. The femur is shortened (as in all types) and a coxa vara deformity is present. The cartilaginous neck is not seen on early radiographs but later ossifies.

Class B: This type shows that the acetabulum is "adequate" or moderately dysplastic and contains the femoral head. No osseous connection is seen between the femoral head and the shaft at maturity. The femoral segment is short and usually has a bulbous bony tuft.

Class C: The acetabulum is severely dysplastic in this type. The femoral head is absent or is very

small and not attached to the femoral shaft. The shortened femoral segment has a tapered proximal end.

Class D: Is the most severe type with absence of the acetabulum and the proximal femur. No proximal tuft is present.

Although it was reported that the above classification does not permit definite classification of PFFD during the first year of life[5, 8], this case presented can still fall under class C despite the fact that the patient is 25 days old. This is evident by the presence of dysplastic acetabuli, absence of femoral heads and shortened femoral shafts which showed some tapering at their proximal ends. However, late ossification may occur whereby portions of the femur may become apparent, had the patient not been lost to follow-up.

Amstutz and Wilson[10] further subdivided Aitken's classification into five types. They divided class A into types 1 and 2. Type 1 is reserved for the milder form with simple femoral shortening and coxa vara. In type 2, a subtrochanteric pseudoarthrosis is present. The remaining types correspond to those of Aitken's classification.

Hip arthrography is also a useful imaging modality in the diagnosis of PFFD but its use involves the risk of ionizing radiation and is invasive in nature[5]. Arthrography was not done in this patient because of the patient's father refused further investigations on his child after the plain radiograph. Moreover, arthrography carries risks of infection and trauma, in addition to the above-mentioned risks associated with arthrography.

Sometimes it may be difficult to correctly classify PFFD using plain radiography before skeletal maturation. But with computed tomography (CT) and 3D-CT reconstruction, non-ossified femoral epiphysis and the connection between epiphysis and shaft may be identified[8]. Computed tomographic-angiographic (CTA) reconstruction can be used to depict the spatial configuration of the pseudarthrosis and the vascular pattern of the extremity in patients with PFFD[2, 8].

The value of measuring the length of the femur in the second trimester on obstetrics ultrasonography as a screening tool for PFFD in-utero has also been reported[8]. Unfortunately, the mother of the patient did not attend ANC during the pregnancy. And even after delivery there was resistance by the father of the patient for ultrasonography of the patient's hip.

The role of magnetic resonance imaging (MRI) in patients with PFFD is to help define the cartilaginous proximal femur and the presence or absence of a cartilaginous connection to the femoral head[5]. Therapeutic decisions are based on the detection of a femoral head and the presence of a connection. Also, the severity of coxa vara, if present, will influence treatment selection. The ability of MRI to clearly depict cartilage is of particular value in this setting. Routine coronal and axial MRI images may be adequate; however, oblique images may be useful in some patients. MRI Arthrography can be an adjunct imaging tool. However, the risk of infection and trauma may limit its use[8]. A possible drawback of MRI consists of the need for general anaesthesia in young children to avoid motion artefacts[5]. MRI was not done in this patient because her parents refused to consent to further investigations and treatment for their child.

One of the goals of classifying children with PFFD is to help determine treatment options[5] which are mostly surgical. Initial surgery is frequently performed before the age of 2 years to minimize psychological trauma to the patient and to allow them to adapt to amputation. Therapy for PFFD is directed towards early and satisfactory ambulation and treatment options include iliofemoral fusion, osteotomy, limb-lengthening procedures and amputation followed by the use of prosthesis. Unfortunately, the patient presented in this case was denied any of the treatment options by her parents who opted for traditional treatment. The patient was later lost to follow-up.

Conclusion

Proximal femoral focal deficiency is a rare skeletal anomaly and rarer is its bilateral variant which occurs in 10-15% of cases. Documentation of this rare congenital disorder in the literature has not been reported in Nigeria, to the best of our

knowledge. Diagnosis of this anomaly can be facilitated by plain radiography.

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