ACR Case Report

Proximal femoral focal deficiency: Case Report

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Abstract

Proximal femoral focal deficiency is a rare congenital skeletal abnormality that involves the femur and acetabulum. It is thought to result from an early disturbance of growing mesenchyme. The condition results in leg length discrepancy due to a shortened and hypoplastic femur.
We report a case of a 4-day old male baby who presented with a history of shortened right lower limb from birth and in whom a plain x-ray of both lower limbs revealed a hypoplastic distal right femur. No other associated congenital anomaly was revealed.
There is a need for early radiologic classification of proximal femoral focal deficiency for surgical planning and treatment.

Keywords*:Proximal femoral focal deficiency, congenital, skeletal*

Introduction

Proximal femoral focal deficiency is an uncommon congenital skeletal defect in which there is a failure in development of the proximal femur and acetabulum of varying degrees.[1,2] It has an estimated incidence of 0.11 to 0.2/10,000 births. The bilateral form is rare occurring in 10 to 15% of cases[3.] It may occur with or without fibula hemimelia, which is a congenital absence of the fibula. [2, 4.]

The following is a case of proximal femoral focal deficiency in a 4-day old child, presented because of its classical radiographic features and rarity.

Case Report

Master A.D, a 4-day old male baby born to a 27-year-old house wife and a 34-year-old trader presented to the General Outpatient Clinic of University of Benin Teaching Hospital (UBTH) having been referred from a private hospital where they gave birth to the baby. The mother complained of the baby having a shortened right lower limb from the hip to the knee since she gave birth to the baby. Pregnancy and delivery were uneventful. The baby is the third child in a monogamous setting. The other siblings are alive and healthy. There is no history of congenital anomaly, sickle cell disease in the family or use of un-prescribed drugs during the pregnancy.

On examination, the patient was healthy looking, conscious and alert and showed normal developmental milestones. The weight was 3.2kg. The cardiovascular, respiratory and genitourinary system examination was normal. The musculoskeletal system examination revealed gross shortening of the proximal part of the right lower limb from the hip to the knee (i.e. the thigh) which was mal-rotated. The right leg i.e. from the knee to the foot appeared normal. The left lower limb and other musculoskeletal system examination were normal. Plain x-ray of both lower limb and pelvis revealed that only a hypoplastic portion of the right distal femur in which the proximal end appeared clubbed was present in association with a poorly developed acetabular roof. There was also a delayed appearance of the epiphyseal ossification centres around the ipsilateral knee joint compared to the other knee (Figure 1). The tibia and fibular were normal bilaterally. The left femur was also normal.

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The abdominal organs appeared normal on ultrasound scan.

An impression of proximal femoral focal deficiency was made and the patient was referred to the orthopaedic surgeon, who informed the parents that the child will need to undergo a surgical operation to correct the leg length discrepancy. This was rejected by the parents who subsequently discharged the child from the hospital against medical advice.

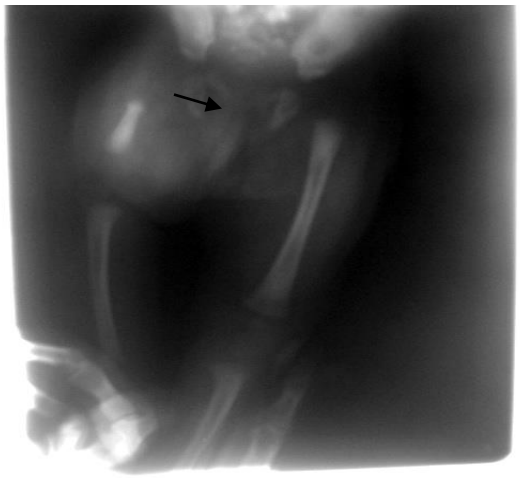

Figure 1: Plain X-ray showing hypoplastic right femur (black arrow) with delayed appearance of epiphyseal ossification around the right knee

Discussion

Proximal femoral focal deficiency is an uncommon defect that involves the femur and acetabulum, as was noted in this case presented, in varying degrees.[1,2] It is often associated with hypoplasia or absence of the fibula and undergrowth of the tibia. Fibula hemimelia occur in about 50% of patients with proximal femoral focal deficiency.[5] The tibia and fibula were however normal in this case presented. Proximal femoral focal deficiency can be unilateral as it was in the case of this patient presented, or bilateral and it is characterised by limb malrotation, deficiency of iliofemoral articulation and limb length discrepancy which was noted in this case presented.[2,6]

Proximal femoral focal deficiency is thought to result from an early disturbance of growing mesenchyme. Boden *et al*[7] carried out a study of the histopathology of the growth plates and epiphyses from a twenty-one-week-old fetus with unilateral proximal femoral focal deficiency and discovered that the growth plate of the proximal part of the involved femur was markedly abnormal in contrast to the growth plates from the distal part of the femur and from all other long bones.

There are different classifications by different authors[1,8,9] but the most popular on the basis of degree of severity and chances of functional restoration is by Aitken[8].

Aitken classification

Type A: The ossification of femoral neck may not be visible in the neonate but eventually ossification occurs. The femoral head develops relatively normally and acetabular roof is well formed. A coxa vara deformity is present.

Type B: A femoral head is present and acetabular roof is reasonably well formed. However, a defect between shaft and neck of the femur persists and hip is unstable. The upper femoral shaft usually is bulbous.

Type C: A femoral head does not develop or is vestigial. The proximal end of the underdeveloped femur is bulbous or clubbed and acetabular roof is poorly developed. The hip is very unstable.

Type D: There is a "pencil sharpened" type deformity of the femur present. There is no femoral head, the acetabular roof is totally dysplastic and the hip is completely unstable.

The patient presented in this case, falls under type C of the above classification.

Plain films will show the shortened femur as the case was in this patient, as well as the commonly associated fibula hypoplasia which was not present in this case or absence and delayed appearance of the ossification centres around the knee when present. The femoral head does not develop as the case was in this patient presented, in severe cases and some indication of whether it is present or not can be inferred by the degree of development of the acetabular roof which was poor in this case on plain radiograph. However, in some cases arthrography is required to demonstrate its presence. In very mild cases ossification between the femoral head and shaft eventually occurs but coxa vara deformity persists. In addition, a few of these cases can develop a pseudoarthrosis through the femoral neck. In its mildest form this condition is referred to as congenital coxa vara. In these cases, the femoral neck is relatively well developed but the femur still is a little

short and a coxa vara deformity is clearly present. In the more severe cases the proximal end of the remaining femoral shaft has a "pencil sharpened" appearance.[1,9] In the case of this patient presented the proximal end was clubbed.

Magnetic resonance imaging can demonstrate the non-ossified cartilaginous femoral capital epiphysis, thus obviating the need for invasive diagnostic procedures and facilitating early classification.[1,10]Pirani *et al*[8] examined the soft tissue anatomy of Aitken types A, B, C and D of proximal femoral focal deficiency using magnetic resonance imaging and a characteristic pattern of soft tissue abnormalities was described. All muscles were present, but most were smaller than their normal counterparts. The exception was obturator externus muscle which was elongated and remained muscular almost up to its insertion. In type A proximal femoral focal deficiency it is straight; in types B, C and D it is L-shaped. The sartorius is hypertrophied. Ultrasonography can be used to make prenatal diagnosis of proximal femoral focal deficiency.[3]

Children with proximal femoral focal deficiency and their families are faced with many treatment decisions, both non-surgical and surgical. Nursing care is central in the care of these children and their families both for psychological support and teaching during the decision-making process so as to help meet post-operative and rehabilitation goals.[2]Treatment objectives include pelvic-femoral stability, prosthetic management, extremity length equality, knee stability and anatomical alignment.[11] There is a need for early radiologic classification for surgical planning and treatment.[10] The milder forms of proximal femoral focal deficiency are quite amenable to therapy but the other types are not. The leg length discrepancy in patients with proximal femoral focal deficiency is difficult to treat and is often unmanageable except by amputation at the level of the ankle joint and subsequent prosthetic fitting in the manner used for above knee amputee.[12]The management of Proximal femoral focal deficiency requires a multidisciplinary team, which includes the pediatric orthopedic surgeon, prosthetists and physical therapists. The goals of treatment are to compensate for the functional deficits. No single treatment approach applies to all cases. Each person with the anomaly must be assessed individually.

In Nigeria and other developing countries where poverty level is high, the people do not generally accept amputation due to sociocultural and economic reasons and thus the disease remains a tragedy for a family that have one.

Summary

A 4-day old baby with proximal femoral focal deficiency is presented. The radiologic diagnosis, classificationand associated anomalies were discussed. The fact that the management of the condition involves a multidisciplinary team was also discussed. The treatment available for this disorder makes it a tragedy for a family with it in developing countries, where high cost of treatment, poverty and lack of adequately trained personnel join together to make prognosis less favourable.

References

1. Anton CG, Applegate KE, Kuivila TE, Wilkes DC. Proximal femoral focal deficiency (PFFD): More than an abnormal hip. SeminMusculoskeletRadiol. 1993; 3(3): 215 – 226.
2. Stormer SV. Proximal femoral focal deficiency OrthopNurs. 1997; 16(5): 25 – 31.
3. Goncalves LF, De Luca GR, Vitorello DA, Parente CM, Philippi H, Saab Neto JA, Pèrèira ET. Prenatal diagnosis of bilateral proximal femoral hypoplasia. Ultrasound Obstet Gynecol. 1996; 8(2): 127 – 30.
4. Sorge G, Arditos, Genuardi M, Pavone V, Rizzo R, Conti G, Neri G, Katz BE, Opitz JM. Proximal Femoral Focal Deficiency (PFFD) and Fibular A/Hypoplasia (FA/H): a model of a developmental field defect. Am J Med Genet. 1995; 55(4): 427 – 32.
5. Ozonoff P. Paediatric orthopaedic Radiology.Philaldelphia, SW Sunders, 1992; 307 – 311.
6. Bryan DD 3rd Epps CH Jr. Proximal femoral focal deficiency: evaluation and management. Orthopaedics. 1991; 14(7): 775 – 84.
7. Boden SD, Fallon MD, Davidson R, Mennuti MT, Kaplan FS. Proximal femoral focal deficiency. Evidence for a defect in proliferation and maturation of chondrocytes. J Bone joint surg Am. 1989; 71(8):1119-29.
8. Pirani S, Beauchamp RD, Li D, Sawatzky B. Soft tissue anatomy of proximal femoral focal deficiency. J pediatrorthop. 1991; 11(5): 563 – 70.
9. Goddard NJ, Hashemi-Nejad A, Fixen JA. Natural history and treatment of instability of the hip in proximal femoral focal deficiency. J pediatrorthop B. 1995; 4(2): 145 – 9.
10. Hillmann JS, Mesgarzadeh M, Revesz G, Bonakdarpour A, Clancy M, Betz RR. Proximal femoral focal deficiency: radiologic analysis of 49 cases. Radiology. 1987; 165(3): 769 – 73.
11. Pappas AM. Congenital abnormalities of the femur and related lower extremity malformations: classification and treatment. J pediatrorthop. 1983; 3(1): 45 – 60.
12. Hamel J, Winkelmann W, Becker W. A new modification of ratationplasty in a patient with proximal femoral focal deficiency Pappas type II. J pediatrorthop B. 1999; 8(3): 200 – 2