CASE REPORT

RIGHT PROXIMAL FEMORAL FOCAL DEFICIENCY (PFFD): CASE REPORT AND REVIEW OF LITERATURE.

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ABSTRACT

Objective: To present a rare case of congenital proximal femoral focal deficiency, Aitken type C in a 6 months old child.

Method/Results: Maternal and family history were evaluated and showed no evidence of diabetes, infection, unprescribed drug use during pregnancy and congenital limb deficiency and neurofibromatosis respectively.

Pelvic and Limb Radiographs showed discrepancies in the lengths of the lower limbs with the right being shorter than the left and relative dysplasia of the right acetabulum.

Conclusion: A radiological disgnosis of congenital PFFD (Proximal Femoral Focal Deficiency) Aitken type C was made.

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INTRODUCTION

Proximal femoral focal deficiency (PFFD), is a congenital anomaly of the pelvis and proximal femur which causes hip deformity and shortening and altered function of the involved lower extremity[1]. The condition may be unilateral or bilateral and is often associated with other congenital anomalies[2]. PFFD consists of failure of normal development of a lesser or greater parts of the proximal femur while some distal femur, by definition, is always present, thus distinguishing it from femoral agenesis[3].
The rarity of this disorder in this environment has prompted the author to report this case.

CASE REPORT

A. K. was a 6 month old child who presented at the University College Hospital, Ibadan with a history of a shortened and deformed right thigh noticed since birth. He was the product of a normal full term pregnancy and he was delivered vaginally. There was no history of maternal diabetes, infection or unprescribed drug usage during pregnancy. The mother VDRL test was negative and the pregnancy was reported uneventful. The family history was also negative for congenital limb deficiency and neurofibromatosis.

Examination of the musculoskeletal system showed a shortened right thigh with prominent skin creases. The right thigh measured 20.5cm and the left measured 29.5cm.
Radiographs of the pelvis and both lower limbs showed absence of the proximal 2/3 of the right femoral shaft with shortening of the femoral shaft and its upper end tapering sharply to a point. There was relative dysplasia of the right acetabulum and the right femoral head was not ossified. Consequently, there was discrepancy in the lengths of the lower limbs with the right being shorter than the left (fig. 1). A radiological diagnosis of congenital PFFD, Aitken type C was made.
Unfortunately the patient failed to keep his subsequent appointment and has been lost to follow up.

Fig. 1

Figure 1: Showing absence of the proximal two-third of the right femoral shaft with shortening of the femoral shaft and its upper end tapering sharply to a point.

There was relative dysplasia of the right acetabulum and the right femoral head was not ossified.

DISCUSSION

PFFD is a congenital limb deficiency involving the proximal end of the femur and the hip joint[4]. The associated clinical problems may include instability of the hip, abduction contractures, malrotation of the femur, weak proximal muscles, formation of pseudar-throsis, hypoplasia of the lateral femoral condyle, and paraxial fibular hemimelia with related deformities[1]. Magdy et al[5] documented that the developing human embryo first shows evidence of limb buds at the 5mm crown-rump stage. As the apical mesoderm proliferates the limb is laid down in a proximo-distal fashion to be completed at the 12mm stage. Elements of the ilium and proximal femur develop from a common cartilaginous anlage, with subsequent cleft formation to create a joint cavity. This means that presence of the acetabulum in a radiograph at any time in the first year of life implies that the femoral head and neck will be present also, even if not evident in the radiograph.

Numerous agents including irradiation, anoxia, ischemia, mechanical or thermal injury, bacterial toxins, viral infection, chemicals and hormones have been postulated causes of PFFD[2]. However, only the drug thalidomide has been showed to be a definitive cause[5]. Thalidomide ingestion 4 to 6 weeks after conception, during the period of limb bud formation and differentiation, produced major limb deformalities[5]. No identifiable etiological factor was seen in this patient

The established classifications of congenital abnormalities of the femur concentrate on detailed examination of radiographs[6]. The best known classifications in the English literature are those of Aitken[7] and Amstutz and Wilson[8]. Aitken's classification concentrates on the presence or absence of cartilaginous continuity of the proximal femur and the development of the hip joint. The classification of Amstutz and Wilson attempts a detailed breakdown of all the anatomical types from a mild degree of femoral hypoplasia to subtotal absence of the femur.

Aitken[9] classified PFFD into 4 types on the basis of radiographic features:

Type (A):

- The femur is short with coax vara and lateral bowing of its upper third.
- There is always adequate acetabulum that contains the femoral head.
- At the subtrochanteric region a pseudoarthrosis develops.
- At the skeletal maturity, ossification of the pseudoarthrosis will take place in most cases, but the varus angulation may be very severe.

Type (B):

- The ossification of the capital femoral epiphysis is delayed and the acetabulum is mildly dysplastic.
- The upper end of the femoral shaft lies above the femoral head.

- The junction between the femoral head and shaft is by defective cartilage that fails to ossify at skeletal maturity.

Type (C):

- The acetabulum is markedly dysplastic and the femoral head never ossify.
- The femoral shaft is very short and its upper end tapers sharply to a point.

  The hip is very unstable.

Type (D):

- Both acetabulum and femoral head are absent.
- The femur is represented by the femoral condyles.

This patient belonged to the type C group

Aitken[9] also documented the associated anomalies found in the patients with PFFD and these included: Congenital anomaly of fibula, the pattela may be absent, small or high riding pattela, lateral subxated or dislocated patellofemoral joint, flexion deformity of the knee (genuvalgum) and unstable knee joint. However, none of these was seen in this patient.

In conclusion patients with a clinical appearance of gross femoral deficiency can be differentiated on clinical grounds into two groups: a milder form, the congenital short femur, where in most cases the goal should be limb length equalization; and a severe form, the true proximal femoral focal deficiency, in which a prosthesis is always necessary and function can be optimized by early knee fusion and tibial rotation plasty[6]. In Nigeria, Adekoya Cole et al[10] documented the use of limb lengthening by distraction osteogenesis in the treatment of limb discrepancy resulting from proximal femoral focal deficiency and conservative management of fractures and epiphyseal injuries in children.

REFERENCES

1. Bryant D and Epps C. Proximal Femoral Focal Deficiency: Evaluation and management. Orthop., 1991; 14:775-783.
2. Koman L. A., Meyer L. C. and Warren F. H.Proximal femoral focal deficiency natural history and management. Clin. Orthop., 1982; 162: 135-140
3. Sutton David: A. textbook of Radiology and Imaging, 1987; Vol.1, 4th Edition. Churchill, Livingstone, Edinburgh; pp 7-9.
4. Eileen F., Ronald Z., Yoshio S. Energy expenditure during walking by children who have proximal femoral focal deficiency. JBJS, 1996; 78 (12): 1857 1862
5. Magdy Abdel Mota'al and Shanmuga Jayakumar. Proximal femoral focal deficiency, November 2003. Available at ttp://gait.aidi.udel.edu/res695/homepage/pd_ortho/educate/Chincase/pffd.htm.
6. Gillespie R. and Torode I. P. Classification and management of congenital abnormalities of the femur. JBJS, 1983 ; 65(5); 557-568.
7. Aitken G.T.Amputation as a treatment for certain lower-extremity congenital abnormalities. JBJS, 1959; 41 A: 1267-85
8. Amstutz H. C. and Wilson P. D Jr. Dysgenesis of the proximal femur (coxa vara) and it is surgical management. JBJS, 1962: 44-A; 1-24.
9. Aitken G.T.Congenital short femur with fibular hemimelia. JBJS, 1974; 56-A: 1306 1309.
10. Adekoya Cole T.O., Adewole O.A., Giwa S.O. and Amabibi A. Limb Lengthening by Distraction Osteogenesis : A Case Report. Nig. Qt. J. Hosp. Med., 1999; 9 (2): 98-99.