

Proximal Femoral Focal Deficiency (PFFD) a Case report.

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Citation

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Abstract

Proximal femoral focal deficiency (PFFD) is a rare but complex problem. It is a non-hereditary birth defect that affects pelvis and the proximal femur. The disorder may affect one side or both, with the hip being deformed and the lower limb shortened. The incidence of deficiency ranges from 1/50000 to 1/200,000 population.

INTRODUCTION

Etiology of PFFD is not known exactly, but certain theories have been proposed and agents implicated. Sclerotome subtraction is one such theory that has been offered to explain several different limb deficiencies. Specifically this theory states that injury to neural crest cells that form the precursors to the peripheral sensory nerves of L4 and L5 result in PFFD.

A second theory, proposed by Boden et al, states that PFFD may be the result of a defect in proliferation and maturation of chondrocytes in the proximal growth plate.

Agents implicated in causing such injuries include anoxia, irradiation, ischaemia, bacterial and viral infection, toxins and hormones. Thalidomide when taken by the mother between the 4th and 6th week of gestation, has been demonstrated to be a definite cause of PFFD.

CASE REPORT

A 4 months old female child was brought to the out-patient department with chief complaint of shortening of the left lower limb. On further evaluation it was found that in addition to shortening of thigh component there was flexion contracture of knee and hip joint of the same side.

There was a true shortening of about 5 centimeters and the shortening was in the femoral component. The gluteal and thigh folds were absent, and the proximal thigh was bulbous.

Figure 1

Figure 1



Figure 2

Figure 2



Figure 3

Figure 3



left femoral epiphysis is smaller in size but contained within acetabulum. The proximal portion of femur is absent on left side. The x-rays of bilateral legs and spine were normal.

Figure 4

Figure 4



Figure 5

Figure 5



RADIOLOGICAL FEATURES

x-ray pelvis with both hips and femurs antero-posterior view is suggestive of normal right femur and acetabulum, while as

Figure 6

Figure 6



DISCUSSION

PFFD is a developmental defect of the proximal femur recognizable at birth. Abnormality ranges from hypoplasia of entire femur to complete absence of the proximal end, bilateral involvement is seen in 15% of cases. In general, in individuals with PFFD, the proximal femur is partially absent, and the entire limb is overall shortened. A few main biomechanical abnormalities are present in children with PFFD, as well as in adults with limb deficiencies. These include limb length discrepancies, malrotation, proximal joint instability, and inadequacy of the proximal musculature.

PFFD can be associated with coxa vera or fibular hemimelia(50%), congenital ligamentous laxity and contractures are also common.

CLASSIFICATION-AITKEN

Class A: femoral head present; severe varus deformity may develop.

Class B: femoral head present, but ossification is delayed; severe varus deformity and pseudoarthrosis may develop.

Class C: femoral head is not present; severe dysplasia of acetabulum; severe shortening of femur.

Class D: femoral head is not present; no acetabulum is present because the lateral pelvic wall is flat; severe shortening of femur.

TREATMENT

Standard orthopedic reconstructive procedures have proved totally ineffective in correcting leg length inequality seen in unilateral PFFD, especially when there is accompanying ipsilateral fibular hemimelia. Treatment must be individualized based on leg length discrepancy, adequacy of proximal musculature, femoral rotation and proximal joint stability.

Treatment options are; limb lengthening or contralateral epiphysiodesis (or both) for mild cases. Iliofemoral fusion, knee fusion, Boyd or Syme amputation or Van Nes rotationoplasty for severe cases.

In summary, treatment is highly individualised and ranges from amputation and prosthetic rehabilitation to limb salvage, lengthening, and hip reconstruction. The natural history of the particular variant and the limitation of surgical reconstruction must be considered.

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