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Proximal Femoral Focal Deficiency – An Uncommon Congenital Anomaly: Case Report and a Review of the Literature

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Abstract **Case Report**

Background: Proximal femoral focal deficiency (PFFD) is a rare congenital skeletal abnormality that may be associated with other bone defects. The incidence varies between 1 case per 50,000 people and 1 case per 200,000 people. PFFD is bilateral in 15% of cases [1]. In people with PFFD, the proximal femur is partially absent, but the distal femoral portion is always present, which serves to distinguish this phenomenon from the entire femoral deficiencies when the whole organs are usually shortened. Identifying these abnormalities on radiographs may aid in the management of these cases because early treatment may help ensure adequate femoral growth. This case report sheds light on this intriguing anomaly, offering an idea of the evaluation of Proximal Femoral Focal Deficiency.

Keywords: proximal femur, bone defects, radiographs, adequate femoral growth.

INTRODUCTION

Proximal femoral focal deficiency (PFFD) is a rare congenital condition that results in limb reduction and disability at a young age. The exact cause of this malformation is unknown and it may present as another level of disease involving the femur next to the acetabulum. Identifying these abnormalities on radiographs may help in better case management of the first treatment center that can help achieve adequate femoral growth.

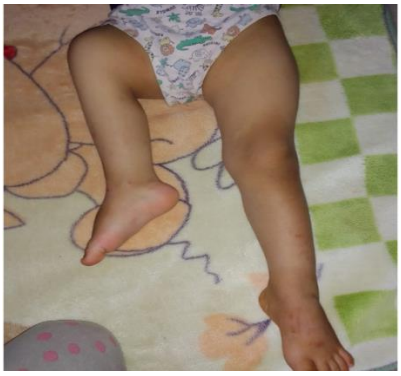
CASE PRESENTATION

In March 2024, a 3-year-old child from our orthopedic department was referred to the radiology department of the same hospital for scannographic evaluation of his lower limbs with inclusion of the pelvis. The main clinical presentation was a history of shortened left lower limb from birth with gradual progression of deformity. There was no history of trauma. Perinatal history was unremarkable. Delivery was uncomplicated, vaginal. Older sibling of the child had no similar complaints.

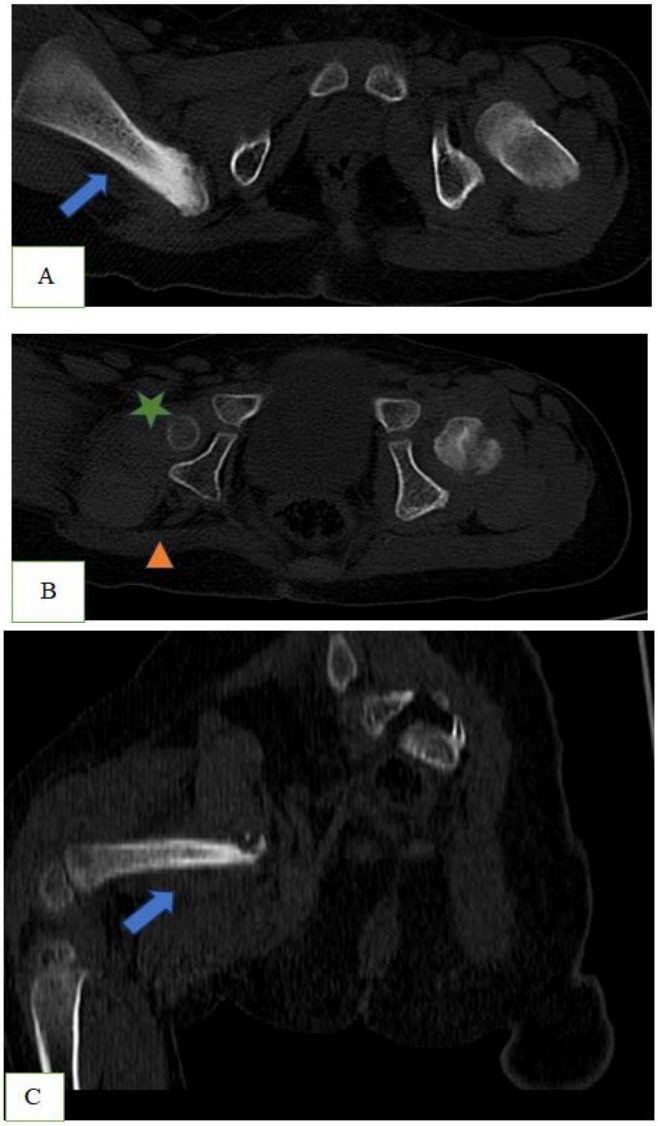
On physical examination, his right thigh was bulky with flexion, lateral rotation, and abduction of the right hip joint, the range of movements at right hip was limited fixed flexion deformity with limitation right knee joint flexion.

Radiographs of his thighs and lower legs revealed a short and hypoplastic right femur and right femoral epiphysis. The shape of his left leg was normal (SCOUT VIEW).

A CT scan of lower limbs was performed and has shown hypoplasia of right femoral bone with mild hypoplastic femoral head and mild hypoplastic right acetabular fossa. The femoral neck is critical shortened and demineralized; based on these findings a diagnosis of proximal femoral focal deficiency class B (classification of Aitken) was made.



Clinical photograph of 3 year old boy with shortened right lower limb and external rotation/abduction of the left thigh



Axial (A, B) and sagittal (C) CT scan images bone and soft tissue window demonstrating hypoplasia of right femoral bone in comparison of the let femoral bone (arrow) with mild hypoplastic femoral head (star)

We can note also the atrophy of right gluteal muscles (triangle)

DISCUSSION

Proximal focal femoral deficiency is a sporadic disease although few familial cases have been described in the literature [2]. It results in non formation of the femoral head with subsequent maldevelopment of the acetabulum [3]. The diagnosis and classification of PFFD have been majorly based on conventional radiographic features.

Key hypotheses postulated are [4]:

- Nutritional deficiency during cell division (4 to 6 weeks of ovulation).
- Mesenchymal tissue damage by local vascular mechanism.
- Gestational femoral compression during diaphyseal ossification.

It is noteworthy that familial predilection has not been recognised.

PFFD manifests itself at birth. Classically femur is shortened with resultant abduction and outside rotation of the worried limb. Flexion contractures of the hip and knee hypoplasia have additionally been reported. Common institutions encompass fibular hemimelia, coxa valga, clubfoot, cleft palate, backbone and cardiovascular anomalies.

There are many classification to describe this malformation but the typically carried out for staging is Aitken's classification, from A to D:

- Class A: Least severe form. Femoral head is present which is attached to the shaft by a cartilaginous neck that ossifies with age. Femoral segment is short. Coxa vara may be present.
- Class B: Femoral head is present. Acetabulum is moderately dysplastic. Absence of osseous connection between the femoral head and shaft.
- Class C: Femoral head is absent. Acetabulum is severely dysplastic. Femoral segment is stunted with a tapered end.
- Class D: Most severe type. Absence of both the acetabulum and proximal femur. Femoral segment is deformed.

Current diagnostic evaluation of PFFD includes plain radiographic examination, bone scintigraphy, ultrasonography and magnetic resonance imaging (MRI). In the younger infant, US may be used for further evaluation of the hip joint. In the older child, arthrography due to its ability to confirm the presence or absence of the femoral head has been used as an adjunctive tool. Cineradiography or push and pull comparison films, as well as abduction-adduction views have also been used to evaluate for hip stability. CT, while useful in evaluating acetabular dysplasia, is generally unhelpful in determining the presence of an unossified femoral head or neck.

More recently, MRI has been used, in conjunction with standard radiographs, for early and more accurate assessment of the articular cartilaginous anatomy and soft tissue prior to ossification of the femoral capital epiphysis. This obivates the need for invasive arthrography and changes in classification with growth as had previously been suggested [5]. This allows earlier treatment planning to benefit treatment outcome.

Treatment of PFFD should be started as soon as possible in order for the femur to grow well. The choice of treatment is made after a thorough evaluation of the bone and soft tissue defects [1]. PFFD class A has little benefit, while classes B, C and D require surgical correction. Alternative treatments available include the use of orthotics, valgus osteotomy, distal femoral epiphysiodesis, Van Nes rotationplasty, Syme amputation and femoral lengthening procedures. These techniques include synchronizing leg length, stabilizing the legs, and achieving pelvofemoral stability.

CONCLUSION

PFFD is a complex congenital anomaly requiring early radiographic classification for operative planning and management. Although plain radiographs remain the mainstay of initial assessment, MRI is useful for early and more accurate assessment of joint cartilage and soft tissue prior to ossification of the femoral epiphysis.

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