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A Case of an Aitken Classification Type C Proximal Femoral Focal Deficiency

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Abstract **Case Report**

Proximal femoral focal deficiency (PFFD) is a rare congenital anomaly characterized by varying degrees of femoral hypoplasia, limb shortening, and hip instability. The exact cause of the disease is not known. We present a case of a 2-year-7-month-old child with unilateral PFFD, evaluated using computed tomography (CT). CT imaging provided detailed visualization of the osseous structures, aiding in classification and management planning. This report underscores the importance of advanced imaging modalities in the assessment and treatment of PFFD. Recognition of this rare abnormality can help manage these cases better since early institution of therapy may help in achieving adequate growth of the femur.

Keywords: Proximal Femoral Focal Deficiency, Aitken Classification, Hip Joint Dysplasia, Congenital Femoral Hypoplasia.

INTRODUCTION

Proximal femoral focal deficiency (PFFD) is a subset of a broader group called congenital femoral deficiency [1]. It is a rare congenital anomaly with an incidence of 1.1–2.0 in 100,000 live births. Most cases of PFFD are unilateral (85–90%) with the right femur is the most frequent culprit [2].

PFFD is a dysplastic phenomenon with a spectrum of femoral involvement. It is defined by deficiency of the iliofemoral articulation, limb malrotation and leg length discrepancy.

The abnormality of the proximal femur ranges from hypoplastic shortened to total absence of proximal two-thirds of femur. Severe affectation may even culminate in femoral agenesis [3]. The condition's etiology is multifactorial, involving genetic and environmental factors during embryonic development.

The diagnosis and classification of this disorder are based mainly on plain radiographs and the relationship between the acetabulum and proximal femur. Aitken's classification is the most common classification scheme, ranging from class A, the least severe, through D. It can be a be applied using computed tomography (CT) imaging as well [4].

Early and accurate diagnosis is crucial for optimal management. While radiographs are commonly used for initial assessment, CT and MRI offer superior delineation of complex anatomical structures, especially in young children where ossification is incomplete [5].

CASE REPORT

A 2-year-7-month-old male child was referred to our pediatric department of radiology for lower limbs CT. The main clinical manifestation was a history of right lower limb shortening from birth and a progressive deformity over time. The pregnancy was uncomplicated and there was no significant family history or prenatal exposure to teratogens. Physical examination revealed a shortened, externally rotated, and flexed right lower limb.

CT imaging of the pelvis and lower limbs revealed demineralized and marked hypoplasia of the right capital femoral epiphysis and flattening of the acetabular roof (loss of concavity) and blunting of the acetabular rim (talus). It also revealed hypoplasia and shortening of the right femur with non-visualization of the femoral neck and metaphyseal junction suggesting absence or severe deficiency of the cervico-metaphyseal segment. The proximal third of the residual femoral diaphysis was tapered, irregular in contour, and shows increased osteosclerosis, indicative of abnormal

ossification and structural remodeling. There was a loss of anatomical alignment between the proximal femoral epiphysis and diaphysis, consistent with postero-medial displacement of the proximal femoral segment (Figure 1).

There was also hypotrophy with fatty degeneration of the muscles of the right hemipelvic girdle and thigh.

These findings are consistent with Aitken classification type C PFFD, characterized by the absence of the femoral head and a severely dysplastic acetabulum.

The left hip joint (left coxo-femoral articulation) was unremarkable, with normal joint congruency and ossification (Figure 2).

Figure 1: Coronal and sagittal CT reconstructions in bone window parameters: marked hypoplasia of the right capital femoral epiphysis and flattening of the acetabular roof (red arrow), hypoplasia and shortening of the right femur with non-visualization of the femoral neck and metaphyseal junction (white arrow), abnormal ossification and structural remodeling of the proximal third of the residual femoral diaphysis (blue arrow)

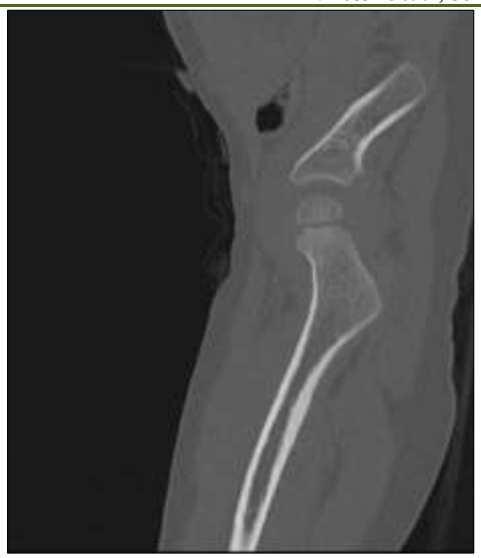

Figure 2: Sagittal CT reconstructions in bone window parameters: normal left coxo-femoral articulation with normal joint congruency and ossification

No other osseous abnormalities were noted. Reconstructive surgery was indicated but deferred until the child is old enough for adequate bone growth and cooperation with rehabilitation.

DISCUSSION

Proximal femoral focal deficiency (PFFD) is a rare, non-hereditary congenital malformation that results in varying degrees of femoral shortening and instability of the hip joint. There is no strong gender predilection. The spectrum of the disease ranges from mild femoral shortening to complete absence of the proximal femur, and may be unilateral or bilateral (bilateral in about 10–15% of cases) [6].

The embryologic basis of PFFD is attributed to a disruption in the mesenchymal development of the proximal femur during the 4th to 8th week of gestation. Teratogenic factors such as viral infections, ischemic events, radiation exposure, and maternal diabetes have been suggested, although the exact etiology remains largely idiopathic [5]. It may be associated with a number of other osseous abnormalities such as fibular hemimelia, absent patella, and acetabular dysplasia [6].

Initial diagnosis of PFFD is typically made via plain radiography using the Aitken classification. This scheme is based upon the presence and location of femoral head and neck on conventional radiography and provides an assessment of future limb function. However, standard X-rays may be limited in young children due to incomplete ossification of the femoral head and acetabulum. In this case, computed tomography (CT) was instrumental in delineating the bony architecture and identifying hypoplasia of the proximal femur, including a deformed femoral head and shortened neck. CT also allows three-dimensional reconstruction, which enhances surgical planning by giving a spatial understanding of femoral displacement and joint integrity.

The Aitken classification [7], revised by Amstutz, is the most widely used to categorize PFFD. It is based on the anatomic relationship between the acetabulum and the proximal end of the femur and designates four classes:

Type a (Least Severe):
- Femoral head is present and attached to the shaft by femoral neck
- A cartilaginous neck may not be seen in early radiograph which ossifies later
- Sometimes, the cartilaginous connection between the neck and shaft may form a subtrochanteric pseudoarthrosis.
- Short femur and coxa vara present.

Type B:
- Acetabulum may be adequate or moderately dysplastic.
- The femoral head is contained in the acetabulum.
- No osseous connection present between the femoral head and shaft at maturity.
- Short femoral segment with a bulbous bony tuft.

Type C:
- Severe acetabular dysplasia.
- Absent or very small femoral head, not attached to the shaft.
- Short femoral segment with a tapered proximal end

Type D (Most Severe):
- Absent acetabulum and proximal femur
- No proximal tuft present.

Amstutz [8], further subdivided Aitken's classification into five types. He 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 pseudarthrosis is present. The remaining types correspond to those of Aitken's classification (Several other classifications were proposed, but Aitken's and Amstutz's classifications are the most widely used.

Since the management of type 1 varies from that of type 2, a distinction between them is necessary.

Our case corresponds to Aitken Type C, characterized by a rudimentary femoral shaft and absent ossified femoral head, with a severely hypoplastic acetabulum. This distinction is critical in prognostication and determining the feasibility of limb-salvage procedures versus prosthetic options [9].

Magnetic resonance imaging (MRI) may also be utilized, especially to assess cartilaginous structures not well visualized on CT, such as the labrum, acetabular cartilage, and unossified femoral head [10]. The use of cross-sectional imaging is invaluable in early childhood where radiographic assessment is limited.

Other conditions can mimic PFFD including congenital short femur (without proximal dysplasia), congenital hip dislocation, femoral hypoplasia–unusual facies syndrome (FH/UFS) and Thalidomide embryopathy.

Proper differentiation using CT/MRI is crucial to avoid misdiagnosis and to guide proper orthopedic referral.

Management of PFFD is complex and highly individualized. Treatment goals include limb length equalization, stability and mobility of the hip and knee, cosmetic acceptability and functional walking ability.

Options range from non-surgical interventions, such as orthotic bracing, to surgical approaches, including rotationplasty, femoral lengthening via the Ilizarov technique, and in severe cases, Van Nes rotationplasty or amputation with prosthetic fitting [11].

Early radiologic identification and classification of PFFD play a pivotal role in directing multidisciplinary management.

CONCLUSION

PFFD, also known as proximal femoral dysgenesis or congenital short femur, is a rare osseous anomaly that may present in isolation or in conjunction with other skeletal abnormalities. This condition leads to limb length discrepancy and gait abnormalities.

Radiographic evaluation is essential for classifying PFFD and assessing its severity. This case highlights the significance of advanced imaging in the evaluation of PFFD. CT provided comprehensive insights into the anatomical abnormalities, facilitating accurate classification and aiding in the formulation of a tailored management plan. Early and precise imaging assessment is vital for optimizing treatment outcomes in children with PFFD.

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