Case Report

Proximal Femoral Focal Deficiency A Case Report
Sagar Jaiswal¹, Chirag Patel¹, Sanjay soni¹, Rajnikant Machhi¹

Abstract

Introduction: Proximal Femoral Focal Deficiency (PFFD) is a rare congenital anomaly, resulting in varying degrees of femoral hypoplasia that causes limb shortening pelvic abnormalities and reduction in the optimal function of the limb. It may be associated with other osseous and systemic anomalies. The exact aetiology is not known and various theories have been put forth. We present a case of an isolated unilateral PFFD that was born locally.

Case Report: A 7 yr old female child was brought to out-patient department at our institute for disability certificate and for opinion with regard to any treatment for the limb anomaly, if any. She was having a short right lower limb, shortened to the level of opposite knee. Detailed history was obtained and she was examined clinically as well as radiologically. She was diagnosed as a case of PFFD. The parents were counselled in detail with regard to the condition and the possible treatment options available. Patients parent refused for any form of treatment for the child.

Conclusion: PFFD is a rare congenital anomaly with variable degree of absence of length of femur. It affects the patient physically, socially as well as psychologically. Proper evaluation and its management accordingly can definitely help the patient to lead a socially and economically productive life.

Keywords: Congenital, limb shortening, femur.
growth plate. Thalidomide has been implicated as a cause when the mother is exposed to it between 4th to 6th weeks of gestation. Other agents which implicated but not proven as a causative factor includes anoxia, ischemia, irradiation, bacterial and viral infections and their toxins hormones, mechanical energy, and differentiated by the presence of limp after weight bearing.

Aitken [3,4] also has stressed that, proximal femoral focal deficiency is a descriptive name for a condition which causes shortening due to a bony deficiency in the femur, in contrast to shortening due to angular or rotatory malalignment.

### Table 1. Aitken classification for PFFD

<table>
<thead>
<tr>
<th>CLASS A</th>
<th>There is normal acetabulum and femoral head with shortening of femur and absence of femoral neck on early radiographs. With age cartilaginous neck ossifies, although this may frequently be associated with a pseudoarthrosis. On healing there is a severe coxa vara with significant limb shortening.</th>
</tr>
</thead>
<tbody>
<tr>
<td>CLASS B</td>
<td>There is a more severe defect which does not heal spontaneously. At maturity there is no connection between the femoral head and the proximal femur; the end of the proximal femur is above the acetabulum. Femoral head though present may have delayed ossification.</td>
</tr>
<tr>
<td>CLASS C</td>
<td>There is further degradation in the formation of the hip, characterized by dysplastic acetabulum, absent femoral head, and short femur. A small, separate ossific tuft can be seen at the proximal end of the femur.</td>
</tr>
<tr>
<td>CLASS D</td>
<td>Here the acetabulum, femoral head, and proximal femur are totally absent. There is no ossified tuft capping the proximal femur. These patient often have bilateral anomalies.</td>
</tr>
</tbody>
</table>

Discussion

PFFD is a rare congenital anomaly of the pelvis and proximal femur with variable degrees of reduction and altered function of the involved extremity; however, a portion of the femur is always present, that differentiates the condition from complete femoral agenesis. The condition is mostly sporadic but a familial form has also been described [14]. The aetiology is as such unknown, but certain theories have been proposed and agents implicated. The sclerotome subtraction theory states that injury to the neural crest cells that form precursors to the peripheral sensory nerves of L4 and L5 results in PFFD [5,9].

Alternative hypothesis states that PFFD is caused by a defect in the proliferation and maturation of the chondrocytes at the growth plate. Thalidomide has been reported. The leg and foot examination virtually unremarkable. Systemic examination including, CNS, CVS, ophthalmological and other musculoskeletal examination including pelvis & spine were unremarkable. Radiological examination (Fig.3) showed near complete absence of right femur with only a remnant of distal femur present. There is an absence of right acetabulum and right fibula. X-ray of other parts of the body were normal. Laboratory data was not significant.

Patients parent were explained in detail about the condition that what exactly the problem is and what the deficiencies their child is having when compared to the normal child of the same age group. We also explained theme about the all possible treatment options available and there their possible consequences. But patients parent refused any form of treatment to their child.

**Figure 1.** Diagramatic Representation of PFFD

**Figure 2.** Clinical Photography

**Figure 3.** Radiological Image
**Conclusion**

Though PFFD is a rare limb anomaly it has great impact on the life the affected patient, which otherwise is normal by growth and development, particularly during their childhood when they grow in peer environment at house, school and social area. Hence timely diagnosis, proper evaluation and accordingly exact management can help the patient to stand themselves capable of living good life. It is of paramount importance to help them by physical and vocational rehabilitation after their surgical treatment.

**Clinical Message**

Management of PFFD poses a great challenge both to the orthopaedic surgeon and family members. The condition has variable presentation. Elaborative evaluation and management is very essential which will have striking effect on the life of patient.

**References**


**Conflict of Interest:** Nil

**Source of Support:** None