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Isolated Unilateral Proximal Focal Femoral Deficiency: A Case Report

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Abstract **Case Report**

Proximal focal femoral deficiency (PFFD) is a rare and intricate congenital disorder, ranging from simple femoral shortening to complete femoral absence. Characterized by inadequate femoral development, we present the case of a 26-year-old patient with a congenital malformation affecting the left lower limb. Clinical examination revealed the patient walked with a pronounced limp due to the shortening of the right lower limb, resulting in a severe limb length discrepancy of 38 cm. An interdisciplinary approach from an early stage is crucial in enhancing functional ability and supporting the social integration of individuals with this condition.

Keywords: Proximal focal femoral deficiency, congenital disorder, prosthesis.

INTRODUCTION

Proximal focal femoral deficiency (PFFD) is a rare congenital malformation that causes insufficient femur development, ranging from femoral shortening to complete absence. This leads to lower limb length discrepancy [1].

Several prosthetic options are available to address the functional impairment caused by this malformation. The excessive cost of these devices can affect their accessibility [2].

CASE PRESENTATION

We present the case of a 26-year-old male patient with a congenital malformation of the left lower limb. The patient's mother reported no exposure to radiation, gestational diabetes, or any history of trauma during pregnancy. Additionally, there is no family history of skeletal abnormalities or other pertinent diseases.

Upon clinical examination, the patient exhibited a significant limp due to a pronounced shortening of the lower right limb, resulting in a severe limb length discrepancy of 38 cm. Notably, his upper limbs and facial appearance were within normal limits.

Standard radiographic imaging revealed femoral hypoplasia, which is classified as stage D according to Aitken's classification. Furthermore, the patient demonstrated pelvic obliquity and rotoscoliosis with a convex curvature to the left, attributable to the limb length discrepancy. [Figure 1,2].

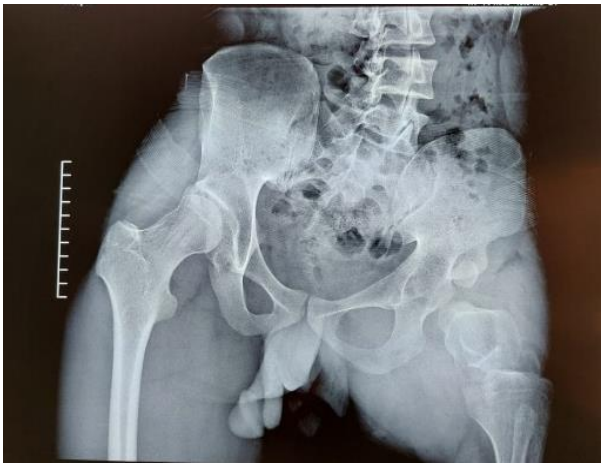

Figure 1: Anterior-posterior view x-ray of pelvis shows a pelvic obliquity, a rotoscoliosis and a shortened left femur

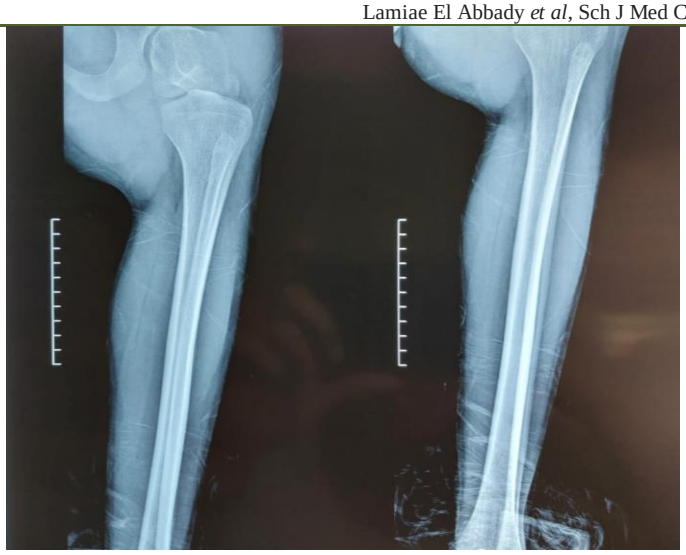

Figure 2: Anteroposterior (A) and lateral (B) tibia-fibula radiographs show a hardly distinguishable tibia bone bending. Note the left crural muscle structures are atrophic in appearance

Our facility provided care that included prescribing a customized prosthesis for the left lower limb and an appropriate rehabilitation protocol [Figure 3,4].

Figure 3, 4: Clinical photos of a proximal focal femoral deficiency patient with a left leg prosthesis from two angles

DISCUSSION

Proximal focal femoral deficiency (PFFD) is a condition characterized by femoral hypoplasia due to a developmental defect in its proximal part. It occurs infrequently, with an incidence rate of 1.1 to 2 per 100,000 live births [2]. The inheritance pattern is typically sporadic, though there have been some reported familial cases [3]. In 85 to 90% of documented instances, PFFD is unilateral, although there are reports of bilateral occurrences [4,5].

Prenatal diagnosis of femoral anomalies is feasible. However, only 19% of cases are identified prenatally, whereas 68% are diagnosed postnatally [5]. Although several factors such as inadequate diabetes management, medication exposure, viral infections, and radiation have been suggested as potential causes of this condition, the precise etiology remains undetermined [5].

PFFD presents with limb length discrepancy and a short thigh positioned in flexion, abduction, and external rotation. The patient showed unilateral shortening of the left lower limb, with the thigh positioned in flexion, abduction, and external rotation. Several classifications of PFFD exist, with Aitken's classification being the most widely used. It differentiates four types of PFFD (A–D) in increasing severity.

This patient's case corresponds to type D, a severe form where the femoral head, neck, and acetabulum are absent. The residual femoral segment is short and deformed. Managing PFFD requires collaboration among orthopedic surgeons, orthotists, and physiotherapists. Management factors include severity, associated deformities, and limb length discrepancy. The therapeutic goals are to restore anatomical alignment, lengthen the affected limb, and stabilize the hip joint [4,9,10].

In cases of 50% shortening or a predicted height discrepancy of less than 20 cm, limb lengthening strategies should be favored. Successful lengthening requires stabilization of the hip and knee joints before any lengthening attempt. If the shortening exceeds 50% or the predicted height discrepancy is greater than 20 cm, as in the case we present, treatment typically involves surgical intervention to facilitate prosthetic fitting. Treatment options include not only prosthetic management but also ankle disarticulation, modified Boyd amputation, knee fusion, rotationplasty, or femoropelvic arthrodesis [11].

Limb lengthening is recommended for cases where there is a shortening of up to 50% or a predicted height discrepancy of less than 20 cm. Stabilize the hip and knee joints before attempting lengthening. When shortening exceeds 50% or the height discrepancy is over 20 cm, surgery is usually needed for prosthetic fitting. Treatment options include prosthetics, ankle disarticulation, modified Boyd amputation, knee fusion, rotationplasty, or femoropelvic arthrodesis [11]. due to the unavailability of limb lengthening facilities or cultural reluctance toward amputation [12,13].

CONCLUSION

Femoral hypoplasia is a rare and complex congenital malformation with highly variable clinical presentations, ranging from femoral shortening to the complete absence of a functional femur and acetabular aplasia. It requires a multidisciplinary and early management approach to enhance functional potential and promote the social integration of patients with such a malformation.

Conflicts of Interest: The authors have no conflicts of interest to declare.

Patient Consent: Written informed consent was obtained from the patient's parents for the publication and accompanying images.

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