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Case Report

Isolated unilateral proximal focal femoral deficiency: A case report and literature review ☆,☆☆

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ABSTRACT

Proximal focal femoral deficiency (PFFD) is a rare congenital anomaly resulting in distinct degrees of femoral hypoplasia and limb shortening. .We present a case of 20-month-old female child, presented with a history of right lower limb shortening from birth and a progressive deformity over time. Physical examination revealed a shortened, externally rotated, and flexed right lower limb. The right and left lower limbs measured 27 cm and 40 cm, respectively. A diagnosis of PFFD was established. Due to limited resources and a shortage of specialized orthopedics doctors 2QA, the patient was referred to a medical mission organization that could help in such cases. Though PFFD is a rare limb anomaly, it significantly impacts a patient's life, if left untreated. Timely diagnosis and early intervention in these cases would have significantly improved prognosis and health outcomes.

Introduction

Proximal Femoral Focal Deficiency (PFFD),also known as congenital proximal femoral deficiency is a rare congenital anomaly with an incidence of 1.1–2 in 100 000 live births [1]. PFFD is marked by hypoplasia of variable parts of the femur, resulting in the shortening of the entire limb and reduction of its optimal function. PFFD usually presents as a shortened, flexed, externally rotated and abducted thigh. It may also occur with or without other congenital anomalies [1–3].

The exact etiology of PFFD is still unknown, however, some risk factors have been shown to cause this anomaly [1,3]. X-ray is an effective diagnostic tool. While MRI is the most recommended method for evaluating PFFD due to its multiplanarity and nonionizing properties [1]. We present here a rare case of a child with isolated unilateral PFFD. This report also provides a comprehensive summary of the PFFD clinical presentation, classification and therapeutic interventions. This work has been reported in line with the SCARE 2023 criteria [4].

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Case presentation

A 20-month-old baby female was referred to the orthopedic department for lower limbs radiographs. The main clinical manifestation was a history of right lower limb shortening from birth and a progressive deformity over time. She was born at 39 weeks of gestation via cesarian section due to obstructed labor. At birth, she weighed 3 kg. Her parents are healthy and have no significant medical history. The pregnancy was uncomplicated and the mother didn't report any abnormalities. Detailed fetal ultrasonography was not performed.

Throughout the pregnancy, she took folic acid regularly and she did not take any non-prescribed medications. The mother reported no radiation, teratogenic agents or hormonal exposure during pregnancy. Also, there was no history of trauma to the mother or the fetus. Her parents are non-consanguineous. There was no family history of similar conditions or other inherited diseases. Her younger sibling is a 3-month-old without any skeletal abnormalities.

Upon physical assessment, the patient appeared in a good general condition. There were no abnormal facial features. Her right thigh was bulky, flexed and externally rotated compared to the left, and her right hip joint was in an abduction position. The right lower limb measured about 27 cm from the anterior superior iliac spine to the medial malleolus, compared to her left lower limb, which measured 40 cm on the same landmarks. There was a 13 cm difference in length between the two limbs (Fig. 1). Her right hip's range of motion was restricted to a continuing flexion deformity whereas the left was normal. Her both upper limbs were normal. Back and spine examination was unremarkable.

A diagnosis of PFFD was suspected. She was referred for radiographic investigation. Plain X-ray of the pelvis and both lower limbs showed shortened and hypoplastic right femur with absent femoral head (Fig. 2). Based on these findings a diagnosis of right unilateral PFFD Class C (Aitken Classification) was made. The patient's parents have been directed to follow the referral process to a medical mission organization that could help her and provide limb-lengthening surgical procedures.

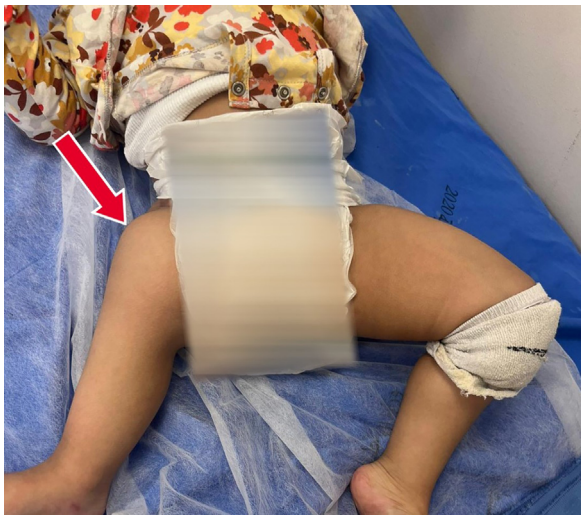

Fig. 1 – Clinical photograph of the patient reveals limb shortening on the right. The right thigh is bulky with flexion, lateral rotation and abduction at the hip (red arrow).

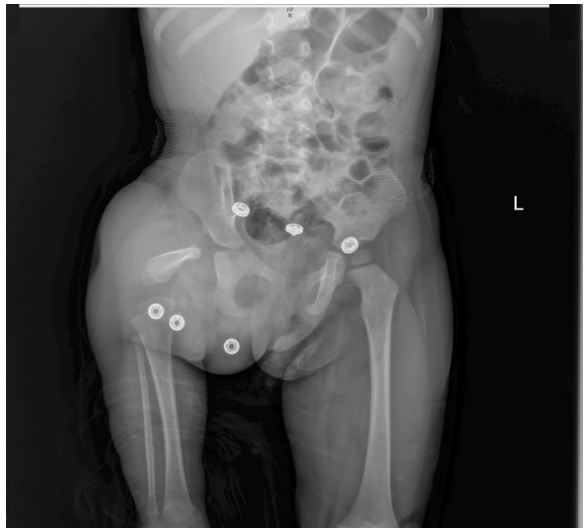

Fig. 2 – Anterior-posterior view x-ray of the lower extremities and pelvis shows a shortened right femur with deficiency of its proximal two-thirds and absent right femoral head.

Discussion

This report mentioned a case of a 20-month-old female child, with rare isolated unilateral proximal femoral focal deficiency (PFFD). PFFD is a subgroup of congenital femoral deficiency, a part of a broader group called congenital limb defect. It's an uncommon condition that is characterized by various degrees of femoral dysplastic changes [5].That might vary from hypoplastic femurs to even a complete absence of the proximal two-thirds of the femur [1,6,7].

PFFD is a sporadic inherited condition, pedigree analysis of this patient identified non-consanguineous healthy parents, with a negative family history. This was consistent with other research showing that PFFD had a sporadic inheritance and no parental age influence [1,7–10]. Most reported cases of PFFD are unilateral, although some publications described bilateral occurrences [1,5,11]. The right femur is the most commonly affected side in unilateral cases (6).

Many risk factors have been linked to PFFD. These include hypoxia, radiation exposure, diabetes mellitus, and thalidomide exposure. Other reported factors are chemical toxicities, hormones, thermal injuries, and trauma to the fetus between the fourth and eighth weeks of gestation [3,5]. It's interesting to note that nothing of these agents was reported neither in the patient nor in her mother. This was consistent with other previously mentioned cases in the literature [1,7,9,10].

The clinical presentation of PFFD may vary widely based on the degree of femoral hypoplasia [1]. Most PFFD cases presented with limb discrepancy and short flexed, abducted, and externally rotated thighs. Other symptoms of PFFD may include Instability of the joints on the affected leg. This patient manifested with right lower limb shortening with flexed, abducted, and externally rotated thigh [1,9].About 30%-60% of PFFD cases were linked to other skeletal anomalies, such as club foot, fibular agenesis, spinal deformity, talipes equinovarus, and, oligodactyly [2,9,12,13]. There was no correlation between her instance and any other congenital anomalies.

Although antenatal diagnosis of PFFD is still a challenge, early detection can offer helpful information about possible management and enable multidisciplinary consultations [6,14]. D'Ambrosio V et al. [2] conducted a review of all prenatally diagnosed PFFD cases between 1990 and 2014. They found that PFFD was suspected in the second trimester in 15 of the 18 cases. Kudla et al. [15] report that a fetus was found to have lower limb asymmetry during the 12th week of gestation through ultrasound examination and was diagnosed with PFFD.In this case, routine prenatal visits show no abnormalities and detailed ultrasonography was not performed.

Prenatal diagnosis or early childhood is the setting for which the majority of PFFD cases that have been reported thus far took place. On the other hand, few PFFD cases were identified in adulthood, Özdemir et al. [16] reported a 20-year-old young woman with isolated unilateral PFFD.

Depending on the conventional radiographic appearance, different authors have described various PFFD schemes. [17–19]. However, the commonly applied classification is Aitken's classification A to D, with class A being the least severe and class D being the most severe. Class A is distinguished by a femoral head with a varus deformity; class B is distinguished by the presence of a femoral head with moderately dysplastic acetabulum, and pseudoarthrosis may occur. Type C lacks both the femoral head and articulation with severely dysplastic acetabulum; and class D is characterized by a femoral head absence combined with severe dysplastic and severely shortened femur [18].This reported case was class C Aitken's classification.

The management of PFFD is indeed challenging and requires multidisciplinary discussion between pediatric orthopedics, prosthetists, and physical therapists [6,7]. Key factors that determine management, include severity, presence of associated deformities, and extremities disparity. The goals of treatment are to achieve normal anatomic alignment, lengthen the affected extremity, and stabilize the hip [6,7,12,18]. If PFFD is neglected or delayed, it can cause serious psychological damage, curvature of the spine leading to chronic back pain, and severe functional disability [7,20].

The severity of limb length discrepancy determines the optimal treatment method. Children with more than 50% shortening or a difference of more than 20 cm usually require ambulation with surgical prostheses and reconstruction surgeries [21]. Surgical procedures may include ankle disarticulation, Boyd amputation, femoral-pelvic arthrodesis, and rotationplasty [1,7,11,21]. A favorable long-term prognosis following surgical orthopedic was found in numerous studies. If the anticipated disparity is less than 20 cm, limb lengthening techniques can be considered To ensure success, stabilization of the hip and knee joints must be established before attempting any lengthening [3,11,14,21].

Palestine, lacks health care facilities for limb-lengthening in addition to the shortage of skilled pediatric orthopedic surgeons [7]. Therefore, the patient was referred to a specialized medical mission organization that could help and offer limb-lengthening surgical procedures.

Conclusion

This report presents a rare congenital anomaly, that significantly impacts a patient's life. Therefore, timely diagnosis, and accordingly appropriate management can help the patient of living a good life. Unfortunately, in facilities with limited resources, the definitive management of this abnormality is demanding, and challenging. It is of paramount importance also to help patients by physical and vocational rehabilitation after their surgical treatment.

Patient consent

Written informed consent was obtained from the patient's parents for publication and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request

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