International Journal of Case Reports in Orthopaedics

E-ISSN: 2707-8353
P-ISSN: 2707-8345
IJCRO 2024; 6(1): 64-67
www.orthocasereports.com
Received: 02-01-2024
Accepted: 03-02-2024

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Proximal focal femoral deficit managed with limb reconstruction system: A rare case report

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DOI: https://doi.org/10.22271/27078345.2024.v6.i1b.191

Abstract
Proximal femoral focal deficit (PFFD) is a rare congenital condition with varying absence of proximal femoral segment associated with shortening of the entire lower limb. We report a case of a 15-year-old female having PFFD with significant shortening of the right limb, and lengthening was achieved through the limb reconstruction system (LRS). Reporting a case of PFFD is a considerable entity due to its rare occurrence. Thus, we report this case in which the patient underwent successful lengthening of the right lower limb. Limb lengthening using the Limb reconstruction system is a successful attempt in patients with limb length discrepancy due to PFFD.

Keywords: Proximal femoral focal deficit, rare congenital condition, Limb reconstruction system

Introduction
Congenital limb defects represent a variety of disorders of cartilage and bone [1]. Congenital femoral insufficiency includes different types of anomalies ranging from the dysplastic hip with a relatively normal femur and shortening of the femur to the most critical condition of total absence of femur [2]. Proximal femoral focal deficit (PFFD), also known by the term congenital proximal femoral deficit (CPFD), is an uncommon congenital skeletal disorder defined as a shortening of the lower extremities due to incomplete absence of the proximal femoral segment [3, 4]. The etiology of PFFD is unknown. However, certain hypotheses describe it as a genetic disorder. The primary ossification center's defect is considered the mainstay pathology [2]. Certain associations with other skeletal conditions like fibular hemimelia, pes equinovarus, oligodactyly, tibial bowing, absence of the cruciate ligament (s), spinal dysraphism, microcephaly, and contralateral upper and lower limbs deformities can also exist [5]. There is variation in incidence from 1 case per 50,000 people to 1 case per 200,000 people [6]. Unilaterality is seen in 60% of the cases, and the remaining 15% to 30% are cases with bilateral proposition. In most unilateral cases, affection is seen in the left lower extremity [6]. The incidence of additional anomalies, such as cleft palate and clubfoot, can be up to 70% [7]. The most common association is with fibular hemimelia [6].

Case report
We present a case of a 15-year-old female. She presented to OPD with complaints of difference in the length of bilateral lower limbs since birth, the right lower limb being shorter than the left (Fig. 1). Limb length discrepancy was found to be progressive in nature. There was no history of trauma, fever, or weight loss. No history was suggestive of childhood affliction of the hip or knee joint. A detailed history suggested an uneventful antenatal period. She was born via normal vaginal delivery as a full-term baby with no postnatal complications. On examination, there was no swelling, scar, sinus, or discharge from the right lower limb. There was no local rise in temperature, no localized tenderness, and the hip range of motion was within normal limits. There was 4.5 cm of supratrochanteric shortening on the affected side.
The radiographs of bilateral lower limbs and pelvis suggested features of PFFD. After a detailed workup, the patient was planned for distal femur corticotomy and Limb Reconstruction System (LRS). Intraoperatively, distal femur metaphyseal osteotomy was done through an anterolateral incision. Three pins each were put in the proximal and distal fragments. Two additional pins were put each in the head of the femur and a lesser trochanter for additional stability (Fig. 2 and Fig. 3).

After the period of 4 days, the distraction of corticotomy was started at the rate of 1mm in 4 divided doses in a day. A total of 45 days of distraction was done (Fig. 4). LRS was left in place for the next 50 days for consolidation of regenerate. (Fig. 5). The operative period was uneventful. The patient had a 30-degree restriction of flexion, which was managed by manipulation under anesthesia. (Fig. 6 and Fig. 7)

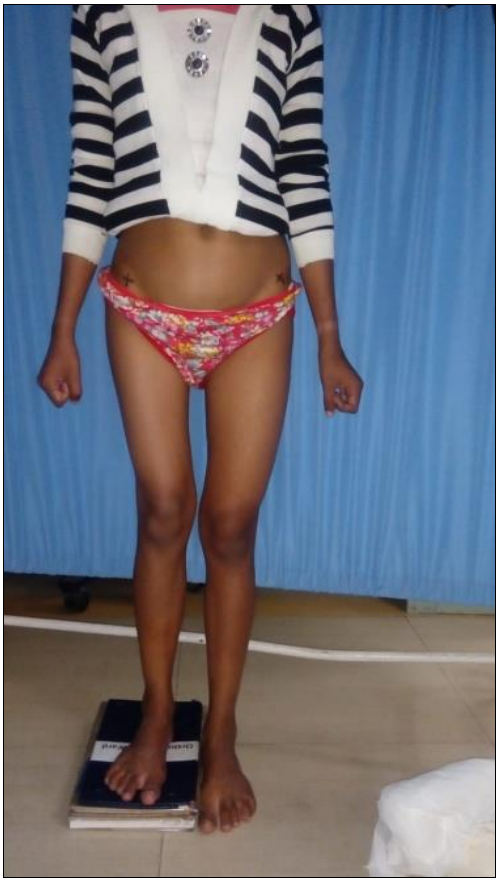


Fig 1: Preoperative clinical image showing both ASIS at the same level with limb length discrepancy.

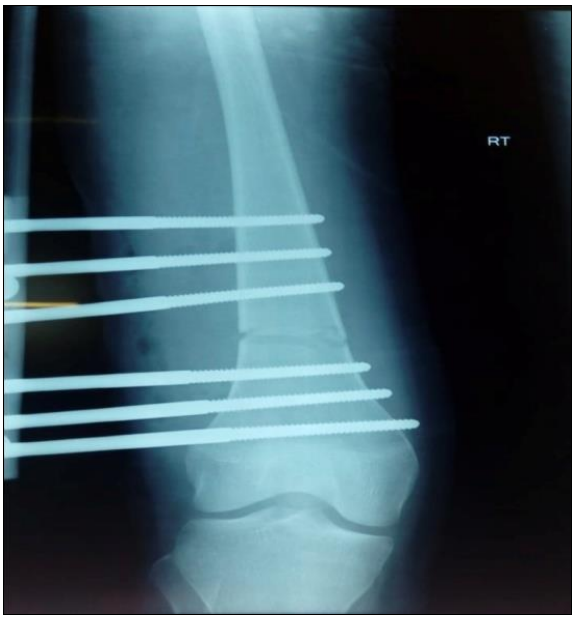


Fig 2: Postoperative X-ray depicting three pins each in the proximal and distal fragment.

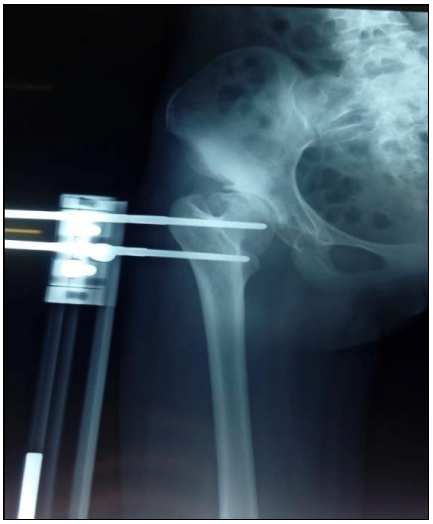


Fig 3: Shows two pins each in the head of the femur and the lesser trochanter.

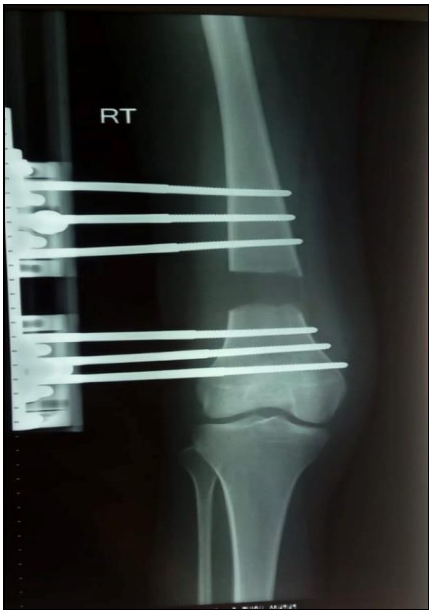


Fig 4: Showing the formation of regenerate post 45 days of distraction

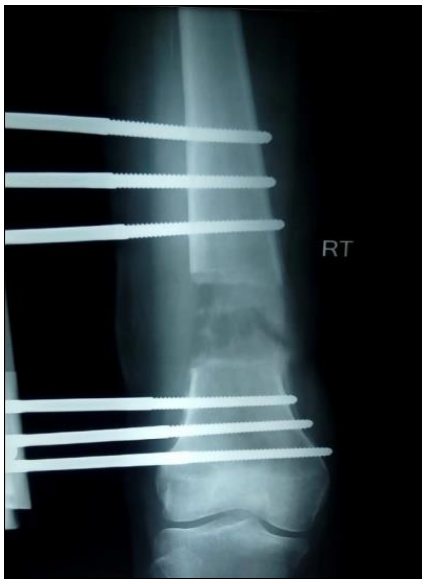
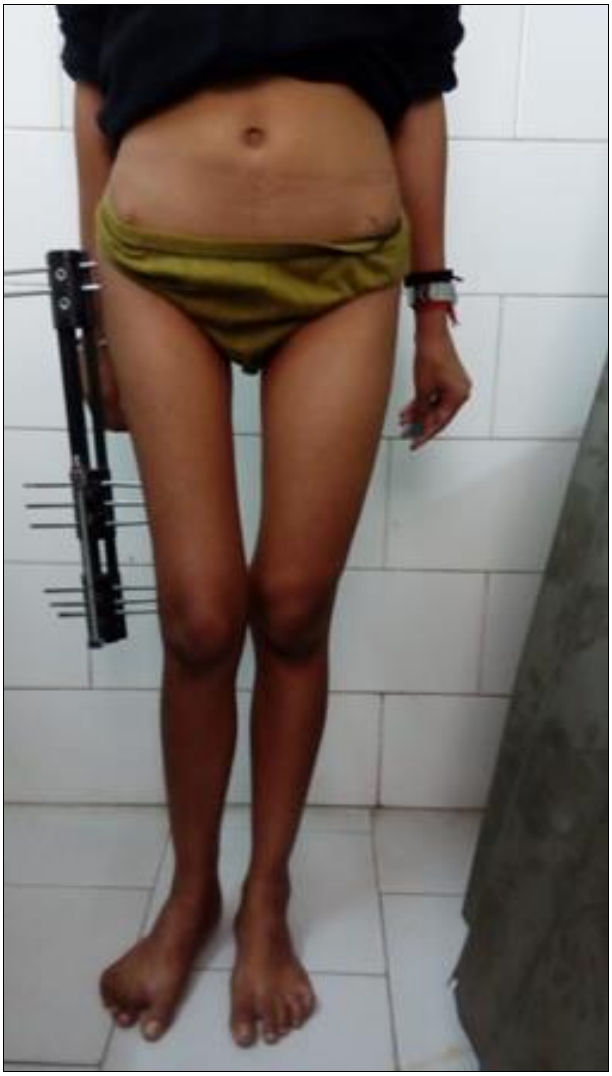


Fig 5: Shows the consolidation of regenerate at four months post-operation



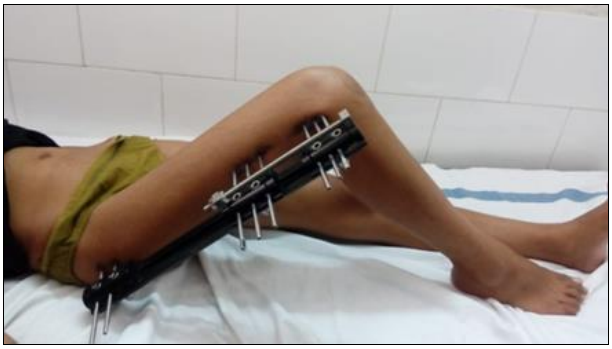


Fig 6 and 7: Showing a clinical postoperative image showing corrected limb length discrepancy with a 30-degree restriction of flexion for which she was taken up for manipulation under anesthesia.

Discussions

As mentioned earlier, PFFD is an extremely rare condition with incidence from 1 case per 50,000 people to 1 case per 200,000 people [6]. That's why our case needs to be included in the existing literature. Risk factors for PFFD are multifactorial and mainly related to antenatal affection of the fetus due to maternal hypoxia, ischemia, poor diabetic control, irradiation, microbiological agents, chemical toxicities, hormones, mechanical or thermal injuries, teratogenic drug intake like thalidomide and trauma to the fetus and/or mother between the fourth to eight weeks of gestation [8]. None of these is linked to our patient or her mother.

On clinical examination, a patient with PFFD shows limb length discrepancy with bulky thigh and lower extremity is seen in the position of flexion, abduction, and external rotation [9]. Our patient had a shortening of 4.5cm with compensatory equines and knee flexion in the affected lower limb.

The commonly used classification used is by Aitken and modified by Amstutz [10, 11]. There are four classes in this classification system; with class a is one in which only the femoral head is involved, having varus deformity; class b has a femoral head with varus deformity and mild acetabular dysplasia; class c with the absence of a femoral head and severe acetabular dysplasia and class d with the absence of both acetabulum and femoral head [10, 11]. Our patient was classified as type A. Our patient presentation was delayed by 15 years of age, so we made the diagnosis and classification using conventional radiography.

The treatment of PFFD is dependent on the amount of shortening in the lower extremity. Lengthening is dependent on discrepancy at maturity. If the discrepancy at maturity is less than 20 cm and the hip and knee joint are stable, then limb lengthening procedures are recommended [4]. This was the scenario in our case where shortening was 4.5cm, and lengthening was done in order to improve the child's ambulatory skills.

Conclusion

The limb reconstruction system can be considered as a successful management strategy for limb lengthening in patients with PFFD. Patients with PFFD can achieve normal gait patterns and carry out their daily living activities post-limb lengthening surgery.

Acknowledgment

Dr. Pravendra Singh analyzed and interpreted patient data and contributed to the writing of the manuscript. Dr. Harsh Kumar performed the clinical examination and contributed to writing the manuscript. Dr. Jashandeep Singh Chahal contributed to the evaluation and management of the patient and the writing of the manuscript. Dr. Palak Gupta contributed to the writing, editing, and submission of the manuscripts. All authors read and approved the final manuscript.

References

1. Nana CT, Ngo-Yamben MA, Fokam P, Naiza M, Nkoke C, *et al*. Proximal Femoral Focal Deficiency (PFFD): A Report of Two Rare Cases in Cameroon. Ortho & Rheum Open Access J. 2021;19(2):556008. DOI: 10.19080/OROAJ.2021.19.556008
2. Boeisa AN, Alarbash MS, Khars AA. Consequences of Proximal Femoral Focal Deficiency Case Report and Literature Review. Int J Ortho Res. 2023;6(3):102-10.
3. Uduma FU, Dim EM, Njeze NR. Proximal femoral focal deficiency–a rare congenital entity: Two case reports and a review of the literature. Journal of Medical Case Reports. 2020 Dec;14(1):1-6.
4. Özdemir M, Kavak RP, Ceylan AH, Cevval ZK. Isolated unilateral proximal focal femoral deficiency presenting in a young woman. BMJ Case Rep. 2020 Jan 21;13(1):e232714.

5. Özdemir M, Kavak RP, Demiral B, Tangobay E. Association of upper limb meromelia, proximal focal femoral deficiency, fibular hemimelia, and intermetatarsal coalition in a young adult male. Radiol Case Rep. 2020 May 1;15(7):882-886.
6. Ukkali SB, Mahvish K, Jeergal N. Congenital femoral deficiency: A rare case report. Int J Contemp Pediatr. 2017 May, 4(3).
7. Ellul M, Chircop M, Grixti CJ, Grech VE. Proximal femoral focal deficiency: a case report.
8. Filly AL, Robnett-Filly B, Filly RA. Syndromes with focal femoral deficiency: Strengths and weaknesses of prenatal sonography. Journal of ultrasound in medicine. 2004 Nov;23(11):1511-1516.
9. Bedoya MA, Chauvin NA, Jaramillo D, Davidson R, Horn BD, Ho-Fung V. Common patterns of congenital lower extremity shortening: diagnosis, classification, and follow-up. Radiographics. 2015 Jul;35(4):1191-207.
10. Aitken GT. Proximal femoral focal deficiency. Limb Development and Deformity: Problems of Evaluation and Rehabilitation. 1969, 456-76.
11. Amstutz HC. The morphology, natural history and treatment of proximal femoral deficiency," in Proximal Femoral Focal Deficiency: A Congenital Anomaly, G T Aitken, Ed., p. 50, National Academy of Sciences, Washington, DC, USA; c1969.

How to Cite This Article
Singh P, Kumar H, Chahal JS, Gupta P. Proximal focal femoral deficit managed with limb reconstruction system: A rare case report. International Journal of Case Reports in Orthopaedics. 2024; 6(1): 64-67.