Rwanda Medical Journal RMJ

CASE REPORT Open access

Proximal femoral focal deficiency associated with fibular hemimelia: an uncommon experience, case report and review of literature

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ABSTRACT

Proximal Femoral Focal Deficiency (PFFD) and Fibular Hemimelia are rare congenital disorders which are among the most common congenital lower limb disorders. PFFD is the absence or shortening of the proximal femur, whereas Fibular Hemimelia is the aplasia or hypoplasia of the fibula. Here we report on a 3-year-old male with clinical and radiological features of PFFD and Fibular Hemimelia. Although this is a rare condition, its occurrence should be considered in daily clinical activities.

Keywords (MeSH): Proximal femoral focal deficiency; Fibular Hemimelia; Congenital lower extremity abnormalities; proximal femoral dysplasia

INTRODUCTION

Proximal Femoral Focal Deficiency (PFFD) is a congenital abnormality of the lower limb which can be as simple as the shortening of the femoral head and neck or as severe as the absence of the acetabulum and proximal femur [1], [2]. It is a rare congenital abnormality of the lower limb whereby there is a significant lower limb discrepancy with an incidence ranging from 1/50,000 up to 1/200,000 [2], [3]. In addition, this congenital defect is often unilateral, between 85% to 90% [4].

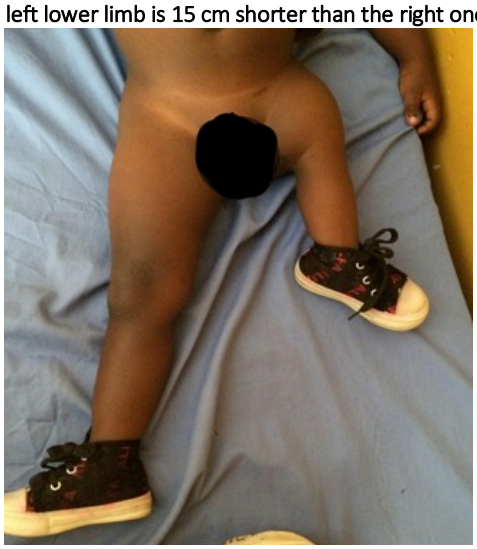
CASE PRESENTATION

In the outpatient department, we received a 3-year-old male presenting with lower limb discrepancy and inability to stand independently. Since birth, the patient's left limb was shorter and as the child developed the limb did not grow like the right leg. The parents also noted that the left hip and buttocks area of the infant musculature were weak and that this weakness was worsening. The patient was born eutocically and cried immediately after birth, with a normal intelligence. The family history is unremarkable. The patient's family eventually reported to have delayed a consultation due to financial reasons.

On examination the left lower limb was found to be 10 cm shorter than the right lower limb (Figure 1), and was held in hip and knee flexion with external rotation. Upon palpation of his hip there was no tenderness, glutei and thigh muscles were weaker compared to the right and there was a decreased range of left hip motion and the baby was unable to extend his left lower limb fully on his own. The rest of his physical examination was normal.

The lower limb radiograph in antero-posterior and lateral views, (Figure 2), led to the diagnosis of left proximal femoral focal deficiency associated with left fibula hemimelia. The patient was not treated because, as we mentioned above, PFFD surgical procedures are very demanding in terms of specialized pediatric centers exacerbated by the financial burden on the family.

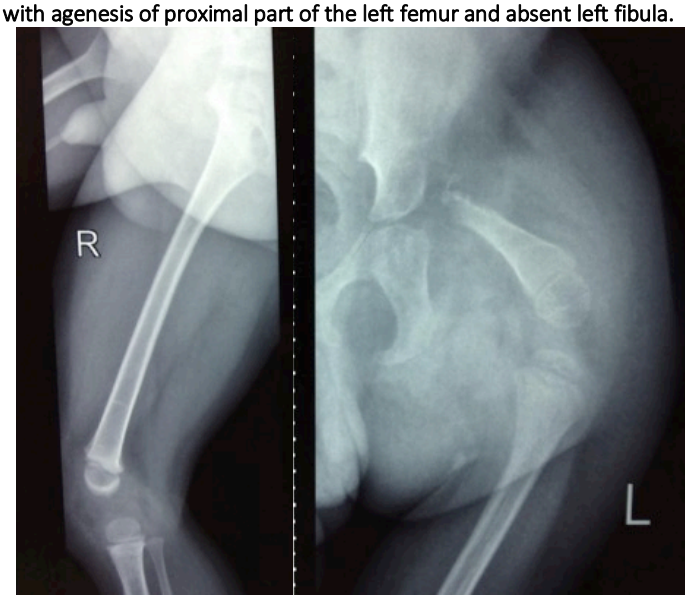
Figure 1: The patient in supine position, there is a discrepancy in limb length. The left lower limb is 15 cm shorter than the right one.



Corresponding author: Prof. Julien GASHEGU kgashegu@gmail.com; **Potential Conflicts of Interest (CoI):** All authors: no potential conflicts of interest disclosed; **Funding**: All authors: no funding was disclosed; **Academic Integrity**. All authors confirm that they have made substantial academic contributions to this manuscript as defined by the ICMJE; **Ethics of human subject participation:** The study not was approved by the local Institutional Review Board as there is no disclosure of patient private information. Informed consent was sought and gained where applicable; **Originality:** All authors: this manuscript is original has not been published elsewhere; **Type-editor**: Sean Batenhorst, USA
Review: This manuscript was peer-reviewed by 3 reviewers in a double-blind review process;
Received: 10th June 2018; **Initial decision given**: 9th July 2018; **Revised manuscript received**: 23rd Sept 2018; **Accepted**: 26th Sept 2018

 Publisher: Rwanda Biomedical Centre (RBC)/Rwanda Health Communication Center, P.O.Box 4586, Kigali. ISSN: 2079-097X
Website: www.rwandamedicaljournal.org

Figure 2: Discrepancy in femur length on right and left femur X-ray, with agenesis of proximal part of the left femur and absent left fibula.



DISCUSSION:

The causes of PFFD are unknown [3], [5], [6], however some theories report an autosomal dominant mutation with possible mosaicism as one of the causes [7]. Moreover,the use of misoprostol in early pregnancy resulted in fetal fibular hemimelia associated with PFFD [8]. In utero injury to neural crest cells, which grow into the L4 and L5 peripheral sensory nerves, and incomplete multiplication and growth of chondrocytes at the responsible growth plate were also stated as possible causes of PFFD [2]. Quite often, PFFD is associated with other birth defects such as: congenital heart disease, spinal cord abnormalities, fibula dimelia, and many others [2], [3], [5], [9]. Jawail S et al. reported a case of PFFD associated with patella and knee cruciate ligaments agenesis [2].Different classifications were elaborated according to the severity of PFFD, the most common being the Aitken classification as shown in Table 1 [10].

Table 1 : Aitken classification of PFFD [10]

Class	Femoral Head	Acetabulum
A	Present	Normal
B	Present	Mildly dysplastic
C	Absent	Severely dysplastic
D	Absent	Absent

Fibular hemimelia, also known as postaxial hypoplasia of a lower extremity, is defined as fibular aplasia or hypoplasia [11], [12]. However, a study done by Sisay S. et al defined Fibular hemimelia as an uncommon congenital fibular absence which can be isolated from or associated with many other abnormalities [13]. In 50% to 80% of cases this congenital defect is associated with PFFD [3]. Among congenital lower limb disorders, PFFD and Fibula Hemimelia are the most common with its diagnosis and classification depending on lower limb radiographic findings [1]. The treatment modalities of PFFD depend on its severity, for limb discrepancy less than 20 cm, nonsurgical management is an option, however, if the discrepancy is greater than 20 cm with severe dysplasia, knee arhrodesis and foot amputation for prosthesis application is an optional choice of treatment [1]. This condition poses a great challenge in terms of management as most of the treatment options focus on making the limb fit for prosthesis. Here we can note Van Nes rotationplasty whereby the limb is rotated so that the ankle joint serves as the knee joint in prosthesis [14]. Patients with mild dysplasia can also benefit from limb lengthening procedures [15]. Here, we report on a Rwandan patient diagnosed with PFFD associated with fibular hemimelia.

Proximal Femoral Focal Deficiency in a association with Fibular Hemimelia is a rare condition as supported by this case report [3]. This toddler first presented at an advanced age while it was show that congenital lower extremity abnormalities can even be seen during antenatal visit by abdominal ultrasonography [5].

This case was similar to those previously described in the literature in that it was unilateral [4]. Our case highlighted PFFD in association with fibular hemimelia which is a similar clinical presentation to that shown by Marthese E, et al [3] reporting PFFD to be often unilateral and 50% to 80% associated with Fibular hemimelia. Moreover the cause of this congenital abnormalities was unknown as supported by a different studies [2], [3], [5], [6].

Conclusion: Proximal femoral focal deficiency associated with fibular hemimelia is an extremely rare condition. However, its occurrence must be kept in mind during antenatal care and in the orthopedic outpatient department and, once found, an exhaustive maternal past medical and obstetric history is paramount as literature supports teratogenicity as a potential cause. In addition, a full physical examination must be performed as these congenital lower limb abnormalities are known to be associated with many other congenital defects. Implementation of a pediatric orthopedic center in resources constrained settings can help in management of those conditions.

REFERENCES

[1] M. a Bedoya, N. a Chauvin, D. Jaramillo, R. Davidson, B. D. Horn, and V. Ho-Fung, “Common Patterns of Congenital Lower Extremity Shortening: Diagnosis, Classification, and Follow-up.,” Radiographics, vol. 35, no. 4, pp. 1191–207, 2015.

[2] S. Jaiswal, P. Chirag, S. Sanjay, and M. Rajnikant, “Proximal Femoral Focal Deficiency A Case Report,” Int. J. Surg. Cases, vol. 1, no. 2, pp. 17–49, 2015.

[3] Marthese Ellul, Marcelle Chircop, Charles Grixti, and Victor Grech, “Proximal femoral focal deficiency - a case report.,” Malta Med. J., vol. 20, no. 02, pp. 42–43, Jun. 2008.

[4] A. K. Agarwal, "Proximal Femoral Focal Deficiency," Jun-2017. [Online]. Available: htt://emedicine.medscape.com.

[5] Marek J Kudla, Aleksandra Beczkowska-Kielek, Katarzyna Kutta, and Justyna Partyka-Lasota, "Proximal femoral focal deficiency of the fetus – early 3D/4D prenatal ultrasound diagnosis.," Med Ultrason, vol. 18, pp. 397–399, 2016.

[6] M. Masaki, H. Kitoh, K. Mishima, Y. Nishida, and N. Ishiguro, "A case of severe proximal focal femoral deficiency with overlapping phenotypes of Al-Awadi-Raas-Rothschild syndrome and Fuhrmann syndrome.," Pediatr Radiol., vol. 44(12), pp. 1617–1619, 2014.

[7] Rabah M. Shawky, Heba Salah Abd Elkhalek, Shaimaa Gad, and Shaimaa Abdelsattar Mohammad, "Unilateral proximal focal femoral deficiency, fibular aplasia, tibial campomelia and oligosyndactyly in an Egyptian child – Probable FFU syndrome," Egypt. J. Med. Hum. Genet., vol. 15, pp. 299–303, 2014.

[8] P. Pallavee, R. Samal, J. Begum, and S. Ghose, "Foetal fibular hemimelia with focal femoral deficiency following prenatal misoprostol use: A case report.," J Obs. Gynaecol., vol. 36(6), pp. 760–761, 2016.

[9] G. Sorge et al., "Proximal femoral focal deficiency (PFFD) and fibular A/hypoplasia (FA/H): a model of a developmental field defect.," Am J Med Genet, pp. 427–432, 1995.

[10] D. Hatch, "Proximal femoral focal deficiency." 2017.

[11] M. C. Holmstrom, "Postaxial Hypoplasia of Lower Extremity (Fibular Hemimelia): Background, Etiology, Epidemiology," Aug-2018. [Online]. Available: https://emedicine.medscape.com/article/1251558-overview#a5.

[12] D. Paley, "Surgical reconstruction for fibular hemimelia," Journal of Children's Orthopaedics, vol. 10, no. 6. pp. 557–583, Dec-2016.

[13] S. Sisay and D. Admassie, "Fibular hemimelia: a rare case report.," Ethiop Med J., vol. 53(3), pp. 159–160, 2015.

[14] J. Ackman et al., "Long-term follow-up of Van Nes rotationplasty in patients with congenital proximal focal femoral deficiency," Bone Jt. J., pp. 192–198, 2013.

[15] D. E. Westberry and J. R. Davids, "Proximal focal femoral deficiency (PFFD): Management options and controversies," HIP Int., vol. 19, no. 1 SUPPL. 6, 2009.