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Case report of congenital proximal femoral deficiency (CPPFD)

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Abstract

Objectives: Congenital Proximal Femoral Deficiency (CPFD), also known as Proximal Femoral Focal Deficiency (PFFD), represents a rare congenital anomaly characterized by partial or complete absence of the proximal segment of the femur, resulting in limb shortening and deformity. The objective of this case report is to highlight the clinical presentation, diagnostic findings, and early management of a rare case of unilateral PFFD in a neonate, emphasizing the importance of prompt diagnosis and multidisciplinary care.

Methods: A 20-day-old male infant was presented with noticeable shortening of the right lower limb and decreased movement at the right hip and knee. Detailed birth and antenatal histories were reviewed to rule out potential teratogenic exposures or familial disorders. Physical examination revealed right femoral shortening and hip dislocation. Radiological evaluation (X-ray and ultrasonography) confirmed proximal femoral deficiency with dislocation and suspected fracture neck of femur. Laboratory investigations, including hematological and biochemical parameters, were within normal limits. Orthopedic consultation suggested card traction for initial management.

Results: The findings were consistent with proximal right femoral deficiency (PFFD), corresponding to Aitken's classification type C. The condition was managed conservatively with card traction, followed by orthopedic assessment for possible future surgical intervention. However, the patient was subsequently lost to follow-up. The case shares similarities with previously documented unilateral right-sided PFFD cases but differs from left-sided presentations reported in other literature.

Conclusions: Early identification and documentation of PFFD are crucial for optimizing functional outcomes. Timely orthopedic evaluation and multidisciplinary management can significantly improve growth potential and quality of life in affected infants. This case underscores the need for awareness and systematic reporting of such rare congenital anomalies.

Keywords: Congenital proximal femoral deficiency, proximal femoral focal deficiency, limb shortening, neonatal orthopedics, case report

Introduction

Proximal femoral focal deficiency (pffd) is a broader group called congenital femoral deficiency (cpfd). (1) Rare congenital anomaly with an incidence of 1.1-2.0 in 100000 live birth. Has a female bias with a male-to-female ratio 1:2^[3] most cases of pffd are u/l (85-90%); rarely b/l (2-3). U/l rt femur is most frequent culprit^[3].

Proximal femoral focal deficiency (PFFD) is a broader group called congenital femoral deficiency (CPFD). (1) It is a rare congenital anomaly with an incidence of 1.1-2.0 per 100,000 live births and shows a female bias, with a male-to-female ratio of 1:2. (3) Most cases of PFFD are unilateral (85-90%), while bilateral involvement is rare^[2-3]. When unilateral, the right femur is the most frequent culprit^[3].

Case Details

A 20-day-old male infant (born of Reetu) presented with complaints of shortening of the right lower limb and decreased movement at the right hip and knee. Birth history: term 40 weeks, birth weight 3 kg, appropriate for gestational age, normal vaginal delivery, cried immediately after birth, delivered at District Hospital Kushinagar. Antenatal history: no history of radiation exposure, known fetotoxic drug exposure, oligohydramnios, trauma, or similar disease in the family. On local examination, there was a short right femur with right

hip dislocation; right thigh measured 11 cm and left thigh 13 cm. Systemic examination was within normal limits. Investigations (X-ray and ultrasonography of both hips and knees) showed a dislocated right hip with shortening of the right femur. Complete blood count, serum electrolytes, C-reactive protein, liver function tests, and kidney function tests were within normal limits. Orthopedic opinion noted proximal femoral deficit with bending, suspected fracture neck of femur (#NOF), and dislocation; card traction was advised.

Clinical Presentation

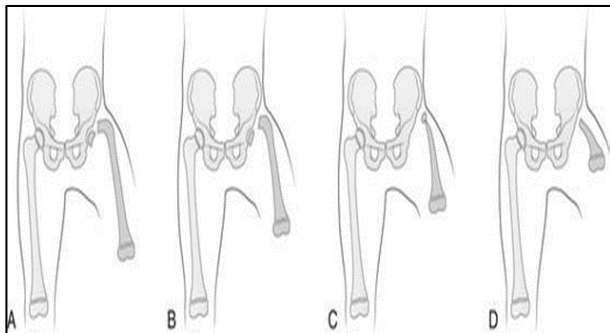


X-Ray



Discussion

- In our case there is a proximal Right femur deficiency seen similar to the case report of *Aditya's p. singh at al* in SMS MC JAIPUR.
- Contrary to the case report of AL-AMEEN MC KARNATAKA by *Sadashiva b ukkali at al* they had found the Left lower limb short i.e PFFD of type C.
- Aitken's classification A to D.
- In our case treatment was given in card traction and further management was in due discussion with ortho department while patients was lost to follow-up due to unknown reason.



Conclusion

We concluded that pffd is a rare congenital anomaly with a spectrum of affection of proximal 2/3rd of the femur. The presence of one newborn case at early staged called for literature documentation.

Besides, such early detection would have warranted timely intervention with consequent anticipated good childhood growth.

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