International Journal of Contemporary Pediatrics
Ukkali SB et al. Int J Contemp Pediatr. 2017 May;4(3):1118-1121
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20171741

Case Report

Congenital femoral deficiency: a rare case report

Sadashiva B. Ukkali, Khodaija Mahvish*, Nazeer Jeergal

Department of Paediatrics, Al-Ameen Medical College Vijayapur, Karnataka, India

Received: 23 February 2017
Accepted: 28 March 2017

***Correspondence:**
Dr. Khodaija Mahvish,
E-mail: dr.mahvish17@gmail.com

ABSTRACT

Proximal femoral focal deficiency (PFFD) is a rare congenital anomaly. We present a case of an isolated unilateral congenital femoral deficiency that was born locally. A full-term male baby was brought to us with a complaint of short left lower limb. Examination revealed short left lower limb with absence of thigh. Detailed history was obtained and he was examined clinically as well as radiologically. He was diagnosed as a case of PFFD of type C. Proper evaluation and its management accordingly can help the patient to lead a socially and economically productive life.

Keywords: Congenital, Femur, Limb shortening, PFFD

INTRODUCTION

Congenital femoral deficiency also known as proximal femoral focal deficiency (PFFD) is an uncommon problem, with an incidence ranging from 1 case per 50,000 population to 1 case per 200,000 population.[1] In general, in individuals with PFFD, the proximal femur is partially absent, and the entire limb is overall shortened. A few main biomechanical abnormalities are present in children with PFFD, as well as in adults with limb deficiencies. Proximal focal femoral deficiency is almost always an isolated occurrence.

However, it may be associated with other skeletal abnormalities like ipsilateral fibular hemimelia (most commonly associated), caudal dysplasia, caudal regression syndrome, lumbosacral spine deformities especially in infants of diabetic mothers.[2]

The aetiology is unknown and no familial or sexual predilection has been recognized.3 Proximal focal femoral deficiency usually is unilateral but is reported to occur bilaterally in as many as 30% of affected individuals. Current understanding is more complete, and various classification systems have been developed. Some postulated that the malformation is due to cellular nutritional disturbance at the time of cell division (at four to six weeks after ovulation). Others postulated a local vascular damage to mesenchymal tissue and some others proposed intrauterine compression of the thigh at time of femoral diaphysis ossification. Heredity does not see to play a major role.[3] Various classification systems have been given but the most widely used is the Aitken classification to provide a systematic taxonomy of this condition.[4,5] We present a case of unilateral isolated PFFD.

CASE REPORT

A full-term male baby deliver by lower segment caesarian section brought to us with a complaint of short left lower limb. The baby was born out of non-consanguineous marriage. Baby was vigorous and hemodynamically stable and breast fed. The course of pregnancy was uneventful. The mother is twenty-four years of age, normoglycemic.

There was no history of radiation exposure, intrauterine infections or any drug intake other than iron, folic acid

and calcium supplements. There is no relevant family history of limb formations.

Physical examination revealed short left lower limb with absence of thigh (Figure 1).

Table 1: Complete blood profile and metabolic screening.

Complete blood profile			Biochemical tests		
Parameters	**Normal values**	**Obtained values**	**Parameters**	**Normal values**	**Obtained values**
White blood cells count (thousands/cumm)	5000-21000	14000	Total bilirubin (mg/dL)	0.10-1.20	5.62
Red blood corpuscles count (millions/cumm)	3.80-5.50	5.26	Alkaline phosphatase (IU/L)	25-100	74
Hemoglobin (g%)	14.50-18.50	17.40	Serum calcium (mg/dL)	8.4-10.4	10.10
Hematocrit (%)	45-56	55	Serum sodium (mmol/L)	135-148	147.90
Mean corpuscular volume (FL)	95-108	106	Serumpotassium (mmol/L)	3.5-5.3	5.24
Mean corpuscular hematocrit (pg)	27-35	33	Serum chloride (mol/L)	98-107	103.50
Mean corpuscular hemoglobin conc. (g/dL)	31-33	31	**Metabolic screening**		
Platelet count (lacs)	2.00-2.50	2.03	G-6-PD (Fluoro immunoassay) (U/gHb)	>2.00	5.10
Neutrophils (%)	60-70	70	Galactosemia (mg/dL)	<8.00	2.30
Lymphocyte (%)	25-30	28	TSH (IU/mL)	<10.00	1.44
Eosinophils (%)	01-04	02	**Microbiological report**		
Monocytes (%)	0-08	00	C- Reactive protein (CRP)	Negative	
Basophils (%)	0-01	00			

G-6-PD: Glucose-6-phosphate dehydrogenase; TSH: Thyroid stimulating hormone.

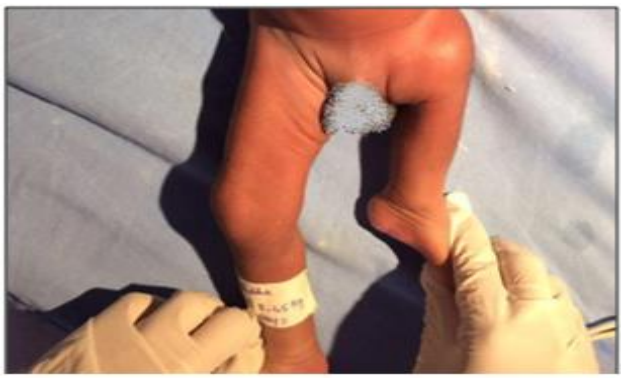

Figure 1: Clinical photography.

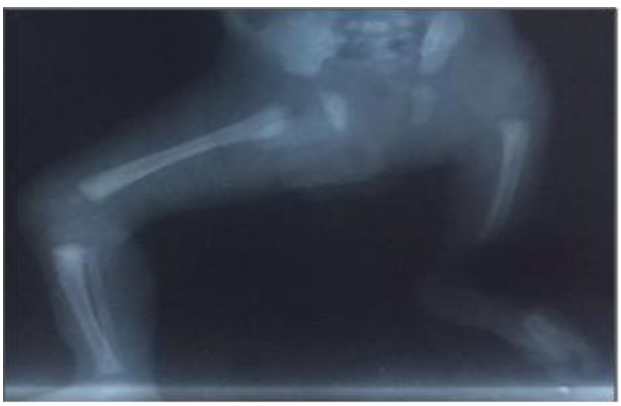

Figure 2: Plain roentgenogram.

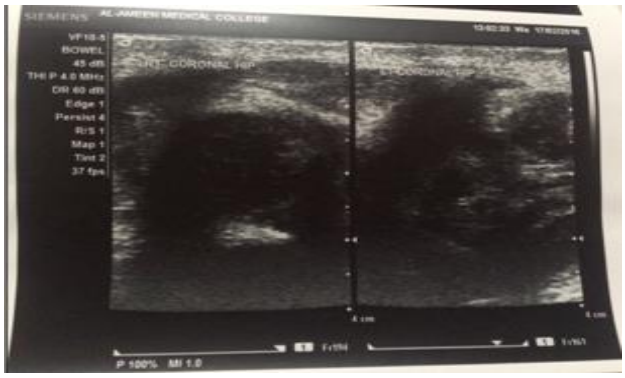

Figure 3: Ultrasonogram.

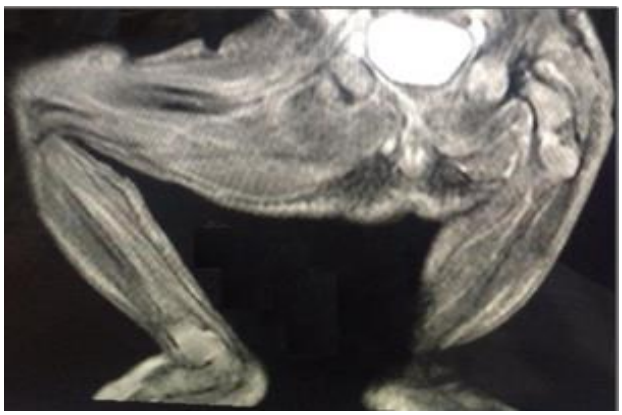

Figure 4: MRI Scan.

In all figures, clinical and radiological examinations was showed.

No other dysmorphic features were noted. Plain roentgenogram showed absence of left femur, while the other parts of skeleton appeared normal (Figure 2). Ultrasonogram of the left hip joint and lower limb showed absence of proximal femur. Ultrasound of abdomen and neurosonogram were normal (Figure 3). Thereafter MRI pelvis and lower limb was done to confirm the diagnosis (Figure 4). It showed evidence of short segmental distal femur with absent proximal femur, head of femur and acetabulum appeared hypoplastic causing shortening, and deformity of left lower limb. The Table 1 revealed complete blood profile, biochemical tests and metabolic screening was within normal limits. The neonate's septic was also negative.

DISCUSSION

Congenital absence of femur is an extreme variant of proximal focal femoral deficiency, a rare congenital anomaly involving lower limbs. Associated bony abnormalities occur with a reported rate of 30% to 60% of which the most common is fibular hemimelia.[6] These anomalies can occur as an isolated defect or in associated with other malformations involving limbs and any other parts of body such as club foot, absence of lateral foot rays, congenital cardiac defects or spinal dysplasia.

Some of the etiological factors include poor diabetic control in early weeks of pregnancy, drug exposure (thalidomide), viral infections, radiation, focal ischemia, chemical toxicity, trauma and causes of familial transmission.[7]

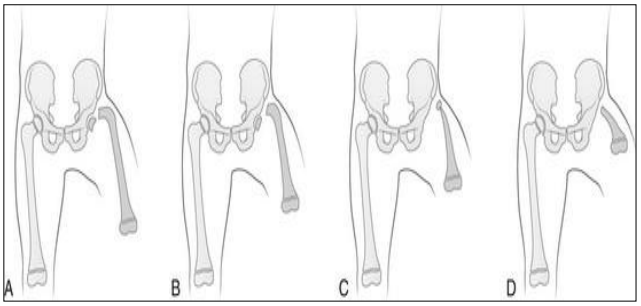

Figure 5: Aitken's classification for proximal femoral focal deficiency.

Congenital femoral deficiency is not usually associated with mental disorders and chromosomal abnormalities. Various differentials in this case includes: Proximal femoral femoral deficiency (PFFD), Focal femoral/ulnar hypoplasia (FFU), Femoral hypoplasia/ unusual facial syndrome (UFS) and Fibular aplasia tibular campomelia-oligosyndactyly (FATCO).[8] In this case, the baby has normal facies and skeletal structure other than femoral hypoplasia, and thus excludes the differentials. Aitken proposed a four-class schema for proximal femoral focal deficiency (PFFD) shown in the following image (Figure 5).[9]

This became the most commonly utilized classification system for PFFD. The Aitken classification is based on the relationship of the acetabulum and the femoral head: (A) Adequate acetabulum, femoral head is present and attached to shaft; (B) adequate acetabulum, femoral head not connected to shaft, which instead has a proximal ossified tuft; (C) dysplastic acetabulum, minimal or absent femoral head, disconnected femoral shaft with tuft; and (D) no acetabulum, no femoral head, shortened proximal femoral shaft with no tuft. Thus, a diagnosis of PFFD type C is made in this case.

CONCLUSION

PFFD patient has great impact on their childhood when they grow in peer environment at house, school and social area. Each person with PFFD must be assessed individually. Current understanding is more complete, and various classification systems have been developed. Hence timely diagnosis, proper evaluation and accordingly exact management can help the patient to stand themselves capable of living good life.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: The study was approved by the Institutional Ethics Committee

REFERENCES

1. Oppenheim WL, Setoguchi Y, Fowler E. Overview and comparison of Syme's amputation and knee fusion with the van Nes rotationplasty procedure in proximal femoral focal deficiency. In: Herring JA, Birch J, eds. The Child With a Limb Deficiency. Chicago: American Academy of Orthopaedic Surgeons; 1998:61-3.
2. Paley D, Chong DY, Prince DE. Congenital femoral deficiency reconstruction and lengthening surgery. In: Pediatric lower limb deformities. New York: Springer International Publishing; 2016:361-425.
3. Chomiak J, Podškubka A, Dungl P, Ošťádal M, Frydrychová M. Cruciate ligaments in proximal femoral focal deficiency: arthroscopic assessment. J Pediatr Orthop. 2012;32(1):21-8.
4. Gillespie R, Torode IP. Classification and management of congenital abnormalities of the femur. J Bone Joint Surg Br. 1983;65(5):557-68.
5. Amstutz HC. The morphology, natural history and treatment of proximal femoral focal deficiencies. In: Aitken GT, ed. Proximal Femoral Focal Deficiency. A Congenital Anomaly. Washington DC: National Academy of Sciences; 1969:1-22.
6. Filly AL, Robnett-Filly B, Filly RA. Syndromes with focal femoral deficiency. J ultrasound Med. 2004;23:1511-6.

7. Kumar D, Krishnamurthy S. A rare association of congenital absence of femur and fibular hemimelia with maternal hyperpyrexia. Indian J Pediatr. 2009;76(11):1178-9.
8. Herring JA. Texas Scottish Rite Hospital for Children. Tachdjian's pediatric orthopaedics. Philadelphia: Saunders; 2008:731-58.
9. Aitken GT. Amputation as a treatment for certain lower extremity congenital abnormalities. J Bone Joint Surg. 1959;41A(7):1267-85.

Cite this article as: Ukkali SB, Mahvish K, Jeergal N. Congenital femoral deficiency: a rare case report. Int J Contemp Pediatr 2017;4:1118-21.