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

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

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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. 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.

The aetiology is unknown and no familial or sexual predilection has been recognized. 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. Various classification systems have been given but the most widely used is the Aitken classification to provide a systematic taxonomy of this condition. 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.

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

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 1). 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. 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.

REFERENCES


