

## *Images in Clinical Practice*

### **Proximal Femoral Focal Deficiency**

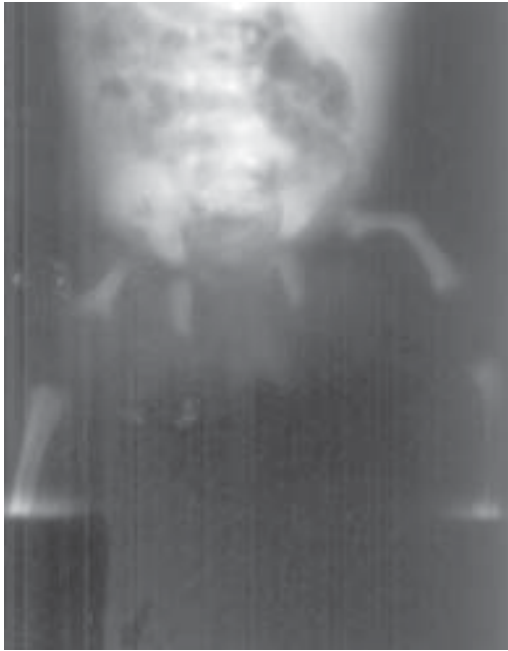
A female baby weighing 1665 g was delivered to a 32 year fourth gravida diabetic mother at 38 weeks gestation by cesarian section. Antenatal ultrasound done at 26 weeks gestation reported bilateral short femur. Examination at birth revealed asymmetrical shortening of lower limbs (left measuring 10 cm, right 9 cm) with talipes equino varus on right and valgus on left (*Fig. 1*). There was no other dysmorphology. Systemic examination was normal. X-ray revealed very short left femur and short curved right femur with

hypoplastic acetabulum (*Fig. 2*). Rest of the skeleton was normal.

Proximal femoral focal deficiency (PFFD) is a rare anomaly with an estimated incidence of <0.2/10,000 births, with bilateral deficiency in 10 to 15% of cases. Nutritional disturbance or vascular insult to the mesenchymal tissue during femoral embryogenesis is postulated as a possible etiology. No known inheritance but recurrence in sibs have been reported. PFFD is almost always an isolated occurrence except for associated ipsilateral fibular hemimelia. It varies in severity from a marginally short femur to a complete absence



*Fig. 1. Shows the baby with PFFD with asymmetrical shortening of lower limbs. Left knee joint (Arrow marked) seen almost at the level of external genitalia.*



*Fig. 2. X-ray showing very short left femur and short curved right femur with hypoplastic acetabulum.*

of femur in severe cases. Kyphomelic dysplasia (rhizomesomelic shortening with lateral bowing of the long bones, flared

irregular metaphyses, a short narrow chest, mild dysmorphic facial features) and campomelic dysplasia (macrocephaly, hypertelorism, long philtrum, micrognathia, hypotonia, hypoplastic scapulae, bowing of the long bones, vertical narrow iliac bones, and absence of ossification of the thoracic pedicles) are important differential diagnosis. Prognosis depends on the degree of hypoplasia. Periodic accurate clinical and radiological examinations are necessary to document the character of the deformities, growth and bone development of femur, and function of the extremity. This will determine the need for surgery and prosthetic supplementation. Prenatal sonography is useful both for detecting PFFD and separating it from syndromes showing global skeletal abnormalities.

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