Radiological Quiz

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Clinical Information

A young boy was brought by his parents with the complaint of very short and bulky left thigh since birth. Radiograph of left side of pelvis, left thigh and left leg is shown.

What is the diagnosis?

Fig. 1 :

(See page 134 for answer).

ANSWER TO THE QUIZ (page 132)

Diagnosis : Proximal Femoral Focal Deficiency

Radiograph shows :

A dysplastic acetabulum

Entire proximal femur represented by a small ossific tuft.

Distal femur represented by only the femoral condyles.

Discussion

Proximal Femoral Focal Deficiency (PFFD) is a rare congenital osseous anomaly characterized by deficiency of a variable portion of the proximal portion of femur [1-3]. It includes a broad spectrum of defects ranging from minor hypoplasia of the proximal femur to almost complete agenesis of the femur [4]. Incidence of PFFD is reported as 1 per 50,000 live births [4]. Exact aetiology of this condition is not known. However maternal diabetes and thalidomide intake by the mother between fourth and sixth week after conception have been attributed as risk factors by various authors [2,4]. A familial link has been described in one case by Schatz and Kopits [1]. PFFD, especially if bilateral, may be associated with other anomalies, like fibular hemimelia, agenesis of cruciate ligaments of the knee, club foot, congenital heart anomalies, spinal deformities and facial dysplasias [4]. Classically patients present with a short bulky thigh resulting in limb length discrepancy. The extremity is abducted, flexed and externally rotated with

variable degree of instability of the proximal joints. Based on radiographic appearance at skeletal maturity Aitken has divided this condition into four classes:

Class A - Normal femoral head and acetabulum with a very short femoral segment and absence of femoral neck on early roentgenogram.

Class B - Same as class A, but with no bony connection between proximal femur and head.

Class C - Marked dysplasia of acetabulum is present. Femoral head does not form, distal femur segment is very short. A small separate ossific tuft can be seen at the proximal end of the distal femur.

Class D - Acetabulum and femoral head are both absent and there is no bony tuft at the proximal end of the deficient femoral shaft [1,4].

In the radiograph shown, there is no evidence of any proximal femoral shaft. The distal femur is represented by the femoral condyles only. The acetabulum, though partially formed, is dysplastic. This is a severe form of PFFD of the class D type, which is rare.

REFERENCES

1. Schatz SL, Kopits SE. Proximal Femoral Focal Deficiency. *Am J Roentgenol* 1978; 131 : 289-95.
2. Epps HC. Current concepts review. Proximal Femoral Focal Deficiency. *J Bone Jt Surg* 1983; 65 A : 867-70.
3. Levinson ED, Ozonoff MB, Royen PM. Proximal Femoral (PFFD). *Radiology* 1977; 125 : 197-203.
4. Crenshaw AH. *Campbell's Operative Orthopaedics*. 8th ed. Vol 3, Mosby 1992; 2117-26.