

## Clinical case

## Proximal focal femoral deficiency complicated by a proximal femoral fracture

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**ABSTRACT.** Proximal focal femoral deficiency (PFFD) is a rare skeletal condition characterized by development failure of the subtrochanteric region of the femoral axis, with several degrees of shortening of the proximal region. There are no case reports of patients with this condition associated with a subtrochanteric fracture. We therefore describe herein the case of a newborn patient with clinical and radiographic diagnosis of proximal focal femoral deficiency complicated by a proximal femur fracture, and managed conservatively with pelvipodalic immobilization, which led to bone healing. The patient will continue to be managed in the long term to assess and treat the sequelae of PFFD according to the growth stage he is in.

**Key words:** fracture, femur, growth and development.

**RESUMEN.** La deficiencia femoral focal proximal (DFFP) es un desorden esquelético raro, caracterizado por la falla en el desarrollo de la región subtrocantérica del eje femoral, con varios grados de acortamiento en su región proximal. No existen reportes de casos de pacientes con esta patología asociada a fractura subtrocantérica, por lo que se presenta el caso de un paciente recién nacido con diagnóstico clínico y radiográfico de deficiencia femoral focal proximal complicada con fractura de fémur proximal y manejada de manera conservadora con inmovilización pelvipodálica con lo que se logró su consolidación. El paciente continuará bajo manejo por un largo plazo para valorar y tratar las secuelas de la DFFP de acuerdo a la etapa de crecimiento en la que se encuentre.

**Palabras clave:** fractura, fémur, crecimiento y desarrollo.

### Level of evidence: IV (Act Ortop Mex, 2011)

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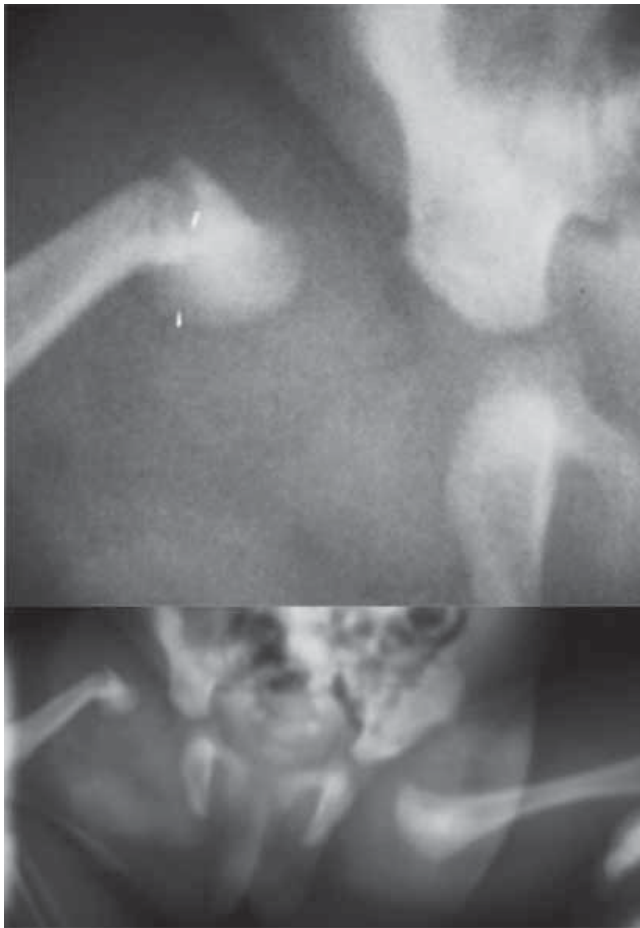
### Introduction

Proximal focal femoral deficiency (PFFD) is a rare skeletal disorder characterized by failure in the development of the subtrochanteric region of the femoral axis, with several grades of shortening of the proximal region.<sup>1</sup> Its incidence rate ranges from 1:50,000 to 1:200,000 live births.<sup>2</sup> The most widely used classification for this condition was developed by Aitken; multiple variants of it have been attempted.<sup>2-4</sup> This condition may be associated with anomalies like ulnar hypoplasia, clubfoot, unusual facies, Pierre Robin syndrome, and it may be a manifestation of Antley-Bixler syndrome.<sup>5</sup> There are reports of multiple syndromes and deformities associated with PFFD. No cases of fractures in patients with this condition have been reported so far.

The purpose of this paper is to report the case of a neonate patient with a diagnosis of proximal focal femoral deficiency complicated by a proximal femoral fracture.

## Clinical case

Female, 6 day-old patient, born through caesarean section after 39 weeks of gestation with pelvic presentation. She is the second daughter of a 31 year-old mother, who received prenatal care, healthy, nurse, with an unremarkable family history. The only remarkable personal history was that during the first weeks of pregnancy she had a migraine attack for which she took tablets containing caffeine and ergotamine once. The father is 32 years old, healthy. She was seen 6 days after birth at the Orthopedics Emergency Room, General Ignacio Zaragoza Regional Hospital, ISSSTE, after being taken by her parents, who reported that she cried intensely when her right pelvic limb was moved; they denied any history of trauma. Pelvic anteroposterior and anteroposterior and lateral X-rays of the right thigh were taken. The diagnosis was subtrochanteric fracture of the right femur and proximal femoral focal hypoplasia (Figure 1). Fracture treatment consisted of a pelvipodal brace and she was followed-up at the outpatient service. Three weeks later, during the control visit, two-plane X-rays were taken, showing appropriate fracture healing and absence of pain upon limb movement.

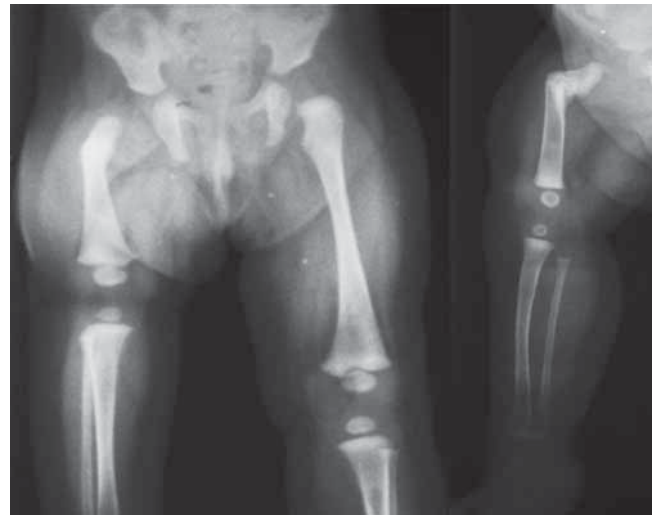


**Figure 1.** Initial X-rays with subtrochanteric fracture and hypoplasia of the right femur. Observe the difference in the development of the right proximal femoral region compared to the left one.

Radiographically, the fracture line had disappeared by week 16 (Figure 2). Physical therapy was started and she continued to be followed-up by the outpatient service to later assess the future complications of proximal focal femoral deficiency and establish a long-term treatment plan (Figure 3).

## Discussion

PFFD is a known congenital entity and, so far, there are no reports of this condition associated with subtrochanteric fracture. In this case the diagnosis of fracture is unquestionable due to the clinical findings in the 6 day-old patient. The evolution of this case allowed observing fracture healing



**Figure 2.** AP and lateral X-rays at week 16. Healed fracture. An important shortening of the right femur is observed compared to the left one.



**Figure 3.** The patient at 4 months of age, with evident shortening of the right thigh.

with conservative treatment, so congenital pseudoarthrosis was ruled out.

Infant abuse has been recognized as a common cause of femoral proximal fractures. In this case the diagnosis may be difficult because the femoral head ossifies after 4 months of age. Other causes of proximal femoral fracture in neonates include obstetric trauma and unintentional trauma. Obstetric trauma is reported as a cause in cases of pelvic presentation or caesarean section.<sup>6</sup> In this case we assume that the fracture occurred at the time of the caesarean section, since the fetus was in a pelvic presentation, and the family denied any type of trauma.

In PFFD there is shortening of the involved limb upon birth, the thigh is very short, the hip is in flexion, abduction and lateral rotation, and the knee in flexion. During subsequent development femoral shortening becomes more evident.<sup>7</sup> Periodic follow-up is therefore very important, as well as proper treatment according to the growth stage the patient is in.

## Conclusion

Conservative treatment is a good option for the management of subtrochanteric fractures in patients with PFFD.

However, it is extremely important to follow-up and manage the underlying condition in the long term to provide the best possible quality of life to our patients with this condition.

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