CONGENITAL BILATERAL SHORT FEMUR COMPLICATED BY STRESS FRACTURE A CASE REPORT

D. KESKİN, N. EZİRMİK, O. KARSAN

Congenital malformation of the femur is an uncommon but complex problem. Short femur with proximal deficiency (Kalamchi type III) is part of this congenital anomaly. If the precautions associated with progressive coxa vara and bowing in the femoral shaft are not taken, stress fractures may occur in the femoral neck and the femoral shaft. We report on a 38-year-old female with type III-A congenital malformation of both femurs who presented the complications mentioned and had not been treated before. This case is instructive because it illustrates the complications developed in patients who have not been treated.

Keywords : congenital malformation ; femur ; complications.
Mots-clés : déficience congénitale ; fémur ; complications.

INTRODUCTION

The spectrum of congenital malformation of the femur ranges from simple hypoplasia (congenital short femur) to complete aplasia (congenital absence of the femur). Congenital defects of the femur can be subdivided into two main categories : those with and those without an osseous defect. Malformation of the femur with an osseous defect of the upper third are generally referred to as proximal focal deficiency. Malformation may also occur in the midportion or distally. The femoral deficiencies without an osseous defect are generally referred to as hypoplasia of the femur or congenital short femur (11).

This congenital limb reduction anomaly was previously discussed under the term "proximal femoral focal deficiency" and was classified according to defects of the proximal segment (1, 3, 8). Afterwards, classification systems including the broad spectrum of congenital changes in the femur were described (7, 10). Kalamchi *et al.* (7) subdivided congenital malformation of the femur into 5 groups (table I).

These patients are usually brought by their parents to the hospital during childhood, and medical treatments are used for this disease. We report the case of a female patient with type III-A congenital

Table I. — Classification of congenital malformation of the femur according to Kalamchi *et al.*

Type I : Short femur with good hip joint.
Type II : Short femur and coxa vara.
Type III : Short femur with proximal deficiency :
A. The defect ossifies in various degrees of varus, forming a bony bridge, causing the patients to function with stable hips (short femur with coxa vara).
B. The defect does not ossify and results in pseudarthrosis.
Type IV : Dysplastic distal femoral segment with no hip joint.
Type V : Total absence of the femur.

Atatürk University, Faculty of Medicine, Department of Orthopedics and Trauma, Erzurum, Turkey.
Correspondence and reprints : D. Keskin, Atatürk Üniversitesi Tıp Fakültesi, Ortopedi ve Travmatoloji Anabilim Dalı, 25240 Erzurum, Turkey.

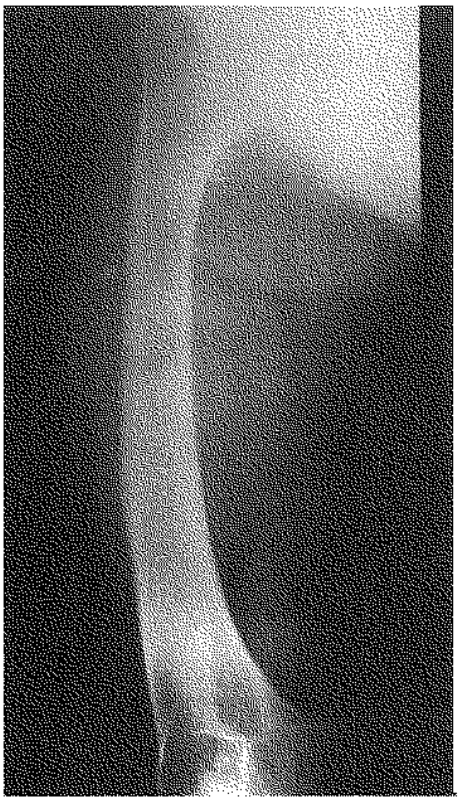

Fig. 1. — Bowing and sclerosis in the right femur

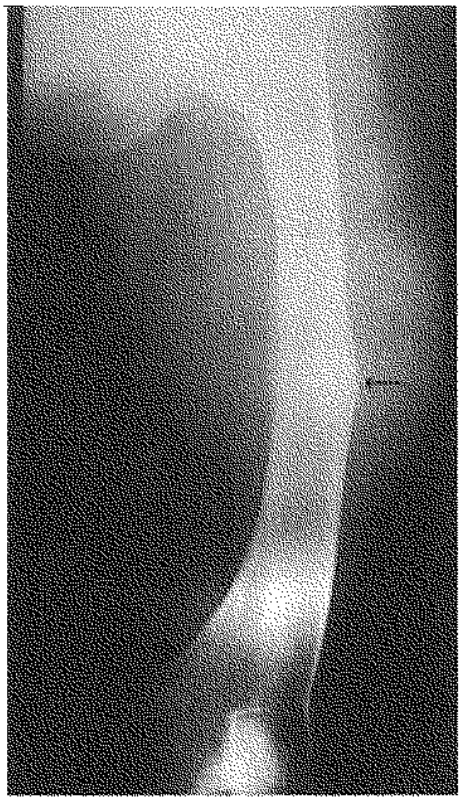

Fig. 2. — Bowing and sclerosis in the left femur, and stress fracture in the lateral cortex of femoral shaft.

deficiency of both femurs who was 38 years old at the time she consulted, and who had never been treated before. This case illustrates the complications which are likely to occur when the necessary treatment has not been carried out in this condition.

CASE REPORT

The patient was a 38-year-old female. Two months before being seen at our clinic, she started complaining of severe pain in her right hip. The pain had been preventing her from walking. On physical examination, there was shortening of both thighs, severe pain was elicited when moving the right hip, and mild valgus deformities were noted in the knees. The examination of other systems was normal. On the radiological examination there were severe coxa vara, bowing and sclerosis in the subtrochanteric and middle regions of the femur and shortening of both femurs (figs. 1, 2). A complete fracture at the base of the right femoral neck and incomplete fractures at the base (proximal and distal cortex) of the left femoral neck and the middle part (lateral cortex) of the left femoral shaft were observed (fig. 3). The other bones and joints were normal.

Prenatal and family histories disclosed no significant findings. The patient had never been treated before. Total hip arthroplasty was suggested for the right hip, but the patient did not accept surgical treatment.

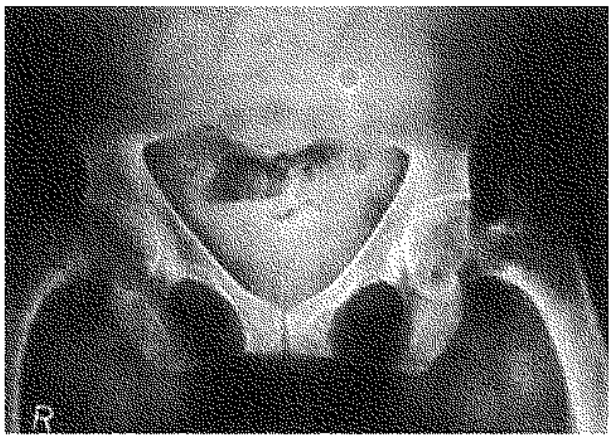

Fig. 3. — Bilateral coxa vara, complete fracture at the base of the right femoral neck and incomplete fracture at the base of the left femoral neck.

DISCUSSION

Congenital malformation of the femur is an uncommon but complex problem. The incidence of associated anomalies is high. Most patients have an additional limb deficiency, but other anomalies such as clubfoot, cleft palate, congenital heart disease, and spinal deformities are rare (4, 6, 7, 10). Our patient had type III-A congenital deficiency of both femurs, however, she did not have other associated anomalies.

These patients are generally brought for medical advice during childhood, and the necessary treatment is begun. The primary objective of treatment is to provide optimal function ; improvement of appearance should always be a secondary consideration. There is no single method of management. Several modalities of treatment are possible, including surgery and fitting of special orthotic devices (2, 5, 7, 9-12).

An important treatment principle for type III is the use of early abduction bracing to prevent excessive varus deformity (7). The treatment objectives for this deformity should include a surgical proposal to correct the biomechanical consequences of the progressive coxa vara. Femoral osteotomies should be performed to correct the neck-shaft angles in cases with excessive coxa vara (5, 7, 10).

Because the treatment and follow-up of these patients are generally started in childhood, it is not clear which complications will develop in the neglected patients. If the precautions related to progressive coxa vara and bowing in the femoral shaft are not taken, there will be stress fractures in the femoral neck and the femoral shaft, as seen in the present case. This case is instructive because it illustrates the complications developed in the patients who have not been treated.

REFERENCES

1. Aitken G. T. Proximal femoral focal deficiency : Definition, classification and management. In : Aitken G. T., ed. *Proximal Femoral Focal Deficiency. A Congenital Anomaly*. National Academy of Sciences, Washington DC, 1969, 1-22.
2. Alman B. A., Krajbich J. I., Hubbard S. Proximal femoral focal deficiency : Results of rotationplasty and Syme amputation. J. Bone Joint Surg., 1995, 77-A, 1876-1882.
3. Amstutz H. C. The morphology, natural history, and treatment of proximal femoral focal deficiency. In : Aitken G. T., ed. *Proximal Femoral Focal Deficiency. A Congenital Anomaly*. National Academy of Sciences, Washington DC, 1969, 50-76.
4. Epps C. H. Current concepts review : Proximal femoral focal deficiency. J. Bone Joint Surg., 1983, 65-A, 867-870.
5. Gillespie R., Torode I. P. Classification and management of congenital abnormalities of the femur. J. Bone Joint Surg., 1983, 65-B, 557-568.
6. Johansson E., Aparisi T. Missing cruciate ligament in congenital short femur. J. Bone Joint Surg., 1983, 65-A, 1109-1115.
7. Kalamchi A., Cowell H. R., Kim K. I. Congenital deficiency of the femur. J. Pediatr. Orthop., 1985, 5, 129-134.
8. King R. E. Some concepts of proximal femoral focal deficiency. In : Aitken G. T., ed. *Proximal Femoral Focal Deficiency. A Congenital Anomaly*. National Academy of Sciences, Washington DC, 1969, 23-49.
9. Kostuik J. P., Gillespie R., Hall J. E., Hubbard S. Van Nes rotational osteotomy for treatment of proximal femoral focal deficiency and congenital short femur. J. Bone Joint Surg., 1975, 57-A, 1039-1046.
10. Pappas A. M. Congenital abnormalities of the femur and related lower extremity malformations : classification and treatment. J. Pediatr. Orthop., 1983, 3, 45-60.
11. Tachdjian M. O. *Pediatric Orthopedics*. WB Saunders Co, Philadelphia, 1990, 553-582.
12. Torode I. P., Gillespie R. Rotationplasty of the lower limb for congenital defects of the femur. J. Bone Joint Surg., 1983, 65-B, 569-573.

SAMENVATTING

D. KESKİN, N. EZİRMİK, O. KARSAN. Congenitaal verkort femur. Een bilateraal geval, verwikkeld met een vermoeidheidsfractuur.

Aangeboren misvormingen van het femur zijn niet alleen zeldzaam maar ook zeer problematisch. Tot deze pathologie behoort ook de congenitale femurverkorting met proximale hypoplasie (type III van Kalamchi). Alhoewel men hier geen voorzorgen hoeft te nemen voor wat betreft een toenemende coxa vara of een femurincurvatie, kan men toch geconfronteerd worden met vermoeidheidsfracturen op het niveau van femurhals en femurschacht. De auteurs beschrijven het geval van een 38-jarige vrouw met een aangeboren misvorming type IIIA van beide femora. De patiënte was steeds onbehandeld gebleven en vertoonde genoemde verwikkeling. Dit geval beschrijft duidelijk hoe therapeutische abstinentie een risico voor verwikkelingen inhoudt.

RÉSUMÉ

D. KESKİN, N. EZİRMİK, O. KARSAN. Fémur court congénital bilatéral compliqué par une fracture de fatigue. Présentation d'un cas.

Les malformations congénitales du fémur sont rares mais représentent un problème complexe. Le fémur court congénital avec déficience proximale (type III de Kalamchi) rentre dans cette anomalie congénitale. En l'absence des précautions qu'imposent une coxa-vara progressive et une incurvation diaphysaire du fémur, des fractures de fatigue peuvent survenir au niveau du col fémoral et de la diaphyse. Les auteurs rapportent le cas d'une femme de 38 ans qui présentait une malformation congénitale de type IIIA des deux fémurs, qui n'avait été soumise à aucun traitement et qui a présenté ce type de complication. Ce cas illustre bien le risque de complication lié à l'absence de traitement.