



## The complications in untreated congenital short femur

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### ARTICLE INFO

### ABSTRACT

#### Article History

Received 27 / 11 / 2013

Accepted 18 / 12 / 2013

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Diagnosis and treatment of patients with congenital short femur is usually dealt with in childhood. It is not, therefore, clear what kind of complications may develop by time when the disease is left to its natural course. In this study, three cases at the ages of 9, 18 and 38 with bilateral congenital short femur, who have never received any treatment before, were presented together with the complications observed. It was determined that in patients who did not receive timely and sufficient treatment, disrupted extremity alignment could affect by time the femoral and the tibial lateral condyles as well as the proximal tibia, causing developmental deficiency and deformations in those locations, and even spontaneous fractures in bone regions where stress was intensified.

*J. Exp. Clin. Med., 2014; 31:247-249*

#### Keywords:

Congenital deformity  
Coxa vara  
Lateral femoral condyle  
Untreated short femur  
Proximal tibial region

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### 1. Introduction

Congenital short femur is a congenital defect presenting itself with a femoral shortness, accompanied sometimes with lateral femoral bowing or coxa vara (Gillespie and Torode, 1983; Pappas, 1983; Kalamchi et al., 1985; Sharrard, 1993; Sanpera and Sparks, 1994; Herring, 2008). Previous studies report that there may be other musculoskeletal defects accompanying to the main findings of this disease (Gillespie and Torode, 1983; Pappas, 1983; Kalamchi et al., 1985; Sharrard, 1993; Herring, 2008). Three bilateral cases in respectively 1<sup>st</sup>, 2<sup>nd</sup> and 4<sup>th</sup> decades, without any previous treatment, were examined in the present study and the relationship between existing deformities and age were investigated.

### 2. Case reports

**Case 1:** A 9-year old girl, who was admitted in our clinic with complaints of limping, and disability of moving the hips outwards. She had received no previous treatment. There were no significant findings in prenatal and family

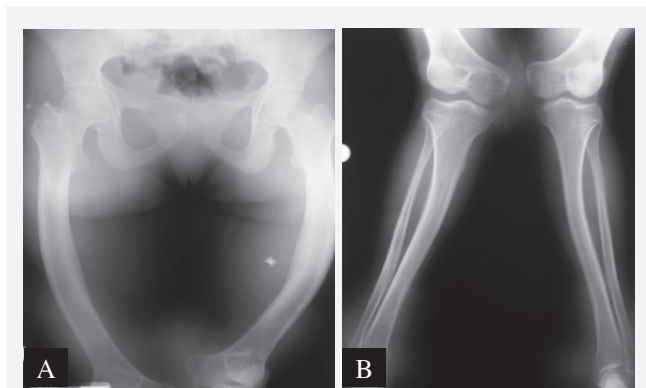
histories. Physical examination revealed increased lumbar lordosis, medium restriction in hip abduction at both sides, shortness in both thighs (more in the right) and angling towards lateral, slight flexion contracture in the right knee and equinus contracture in the right ankle. She walks by stepping over her right fingertips, with trendelenburg gait pattern. Plain radiographs indicated, with the right side being more significant, shortness in both femurs, coxa vara, cortical thickening with narrowing of the medullary canal in the subtrochanteric region, and lateral bowing. In addition, femoral lateral condyles were slightly dysplastic (Fig. 1).

**Case 2:** An 18-year-old female, who was admitted in our clinic with complaints of bend in her knees, severe pain and consequent disability of walking. She had no treatment previously. There were no significant findings in prenatal and family histories. Physical examination indicated shortness in both thighs, angling towards lateral and excess valgus deformity in both knees, with painful knee motions.



**Fig. 1.** Radiograph of first case shows; bilateral coxa vara, femoral shortening, deficiency of the femoral lateral condyles, cortical thickening with narrowing of medullary canal, and lateral bowing in the subtrochanteric femoral regions.

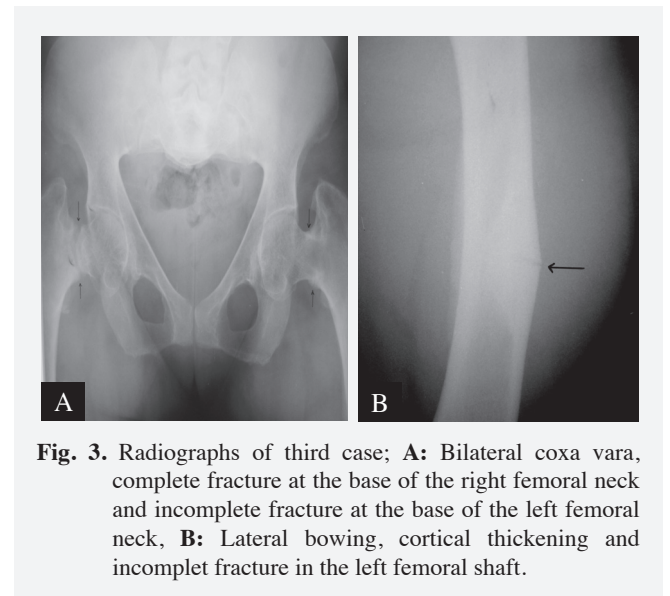
Plain radiographs revealed shortness in both femurs, cortical thickening starting from middle of the shaft extending towards trochanteric region, medullary narrowing and severe lateral bowing. Femoral lateral condyles were severely dysplastic. At the connection junction of both tibias at 1/3 medium and 1/3 proximal, there was an indication of bowing towards medial as well as dysplasia in tibial lateral condyles (Fig. 2A, B).



**Fig. 2.** Radiographs of second case; **A:** Bilateral femoral shortening and lateral bowing in midshaft of the both femurs, **B:** Bilateral dysplasia of the femoral and tibial lateral condyles, and bowing towards medial in the proximal tibial regions.

**Case 3:** 38-years old female, who was admitted in our clinic with complaints of difficulty in walking due to severe pain in her right hip. The pain had started two months ago with no apparent reason. She had no treatment previously.

There were no significant attributes in prenatal and family histories. Physical examination revealed shortness in both thighs and an angling towards lateral; with extreme pain in the movements of the right hip and medium degree valgus deformity in both knees. Radiological examination showed severe coxa vara, lateral bowing and cortical thickening in the subtrochanteric and middle regions of the femurs and shortening of both femurs. A complete fracture was observed at the base of the right femoral neck while an incomplete one at the base of the left femoral neck and in the middle of the shaft (Fig. 3A, B).



**Fig. 3.** Radiographs of third case; **A:** Bilateral coxa vara, complete fracture at the base of the right femoral neck and incomplete fracture at the base of the left femoral neck, **B:** Lateral bowing, cortical thickening and incomplete fracture in the left femoral shaft.

### 3. Discussion

Congenital short femur is a congenital deformity that arises from ossification defect in the region located between 2/3 lower section and proximal part of the femur shaft, which may sometimes present itself by inclination of the femoral shaft from defect localization towards lateral, as well as presence of a coxa vara (Gillespie and Torode, 1983; Pappas, 1983; Kalamchi et al., 1985; Sharrard, 1993; Sanpera and Sparks, 1994; Herring, 2008). It has been reported that this deformity may be accompanied by congenital absence of cruciate ligaments, by shortness of the tibia and fibula, genu valgum, and by deficiency of lateral femoral condyle development (Gillespie and Torode, 1983; Johansson and Aparisi, 1983; Pappas, 1983; Kalamchi et al., 1985; Sharrard, 1993; Herring, 2008). Using abduction braces to prevent excessive varus deformity in early childhood period and femoral osteotomies in later ages; femoral lengthening procedures in order to correct limb inequality in unilateral lesions or performing epiphysiodesis on the healthy side are among the most commonly employed treatment methods (Epps, 1983; Gillespie and Torode, 1983; Pappas, 1983; Kalamchi et al., 1985; Sharrard, 1993; Bowen et al., 2001; Hosalkar et al., 2003; Herring, 2008). What happens if these treatment methods cannot be applied timely and sufficiently? We can answer this question by looking into the findings obtained from our 3 cases, which were in different age groups with no previous treatment. The congenital defect in the femoral shaft becomes ossified by time, causing shortness, lateral bowing and coxa vara. Lateral bowing and coxa vara in untreated patients may heal by time or, in contrast, they may get worse

and may create negative forces on femoral lateral condyle and tibia that may affect adversely their development.

In our first case, lateral condyle deficiency can be clearly observed in addition to the lateral bowing and advanced state of coxa vara of both femur. In our second case, who is 9 years older than the first case, the deficiency in femoral lateral condyles is much more severe. In addition to the deficiency in femoral lateral condyles, we can observe the deficiency in tibial lateral condyles, a high degree of genu valgum deformity and bowing in the proximal tibias towards medial. This shows that, in patients remained untreated, the leg has a tendency to incline towards outside from the knee in order to reensure extremity alignment disrupted due to

lateral femoral bowing, which may possibly cause pressure on lateral femoral and even on the lateral tibial condyles resulting in deficiencies in these locations, and sometimes in deformations that will angle from the proximal of tibia towards medial. In adult untreated patients, as in our third case, spontaneous fractures may develop in bone regions where the stress is intense (such as base of femoral neck, convex apex of lateral femoral bowing).

I believe that this paper will provide important contributions to existing knowledge of congenital short femur, by showing clearly what outcomes may be obtained upon neglect of congenital short femur patients, who should generally be treated appropriately during childhood.

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