High Prevalence of Coxa Vara in Patients With Severe Osteogenesis Imperfecta

Mehdi Aarabi, MD, Frank Rauch, MD, Reggie C. Hamdy, MD, and François Fassier, MD, FRCSC

Abstract: The purpose of this study was to determine the incidence and clinical presentation of coxa vara in 283 patients with osteogenesis imperfecta (OI). The charts and x-rays of 150 girls and 133 boys with OI were reviewed. The patients were classified according to the Silene classification modified by Glorieux: 94 type I, 90 type IV, 67 type III, 18 type V, 10 type VI, and 4 type VII. The mean age was 9.4 years (range 0.3–23.3). Twenty-nine patients (10.2%) had coxa vara (23 left and 20 right). Fifty-five percent of them were type III, 24% type IV, 13.8% type VI, and 3.4% each of types V and VII. The incidence of coxa vara was 6% in type V, 8% in type IV, 24% in type III, 25% in type VII, and 40% in type VI (P < 0.001 for difference between types I, III, and IV). The mean neck–shaft angle was 99 degrees (range 80–110 degrees), the average head–shaft angle was 104 degrees (range 90–120 degrees), and the mean Hilgenreiner–epiphysial angle was 68 degrees (range 40–90 degrees). Twenty-five patients (36 hips) had previous femoral rodding before diagnosis and seven hips (all type III) had no history of rodding. Abduction and internal rotation of the hip joints were restricted in all patients with this deformity. All children with coxa vara had a Trendelenburg gait. In conclusion, coxa vara in OI is not rare, especially in severe forms of the disease. Regular clinical and radiologic follow-up is indicated in children with previous femoral rodding and in severely affected children, particularly those with type II.

Key Words: coxa vara, osteogenesis imperfecta

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METHODS

This was a retrospective evaluation of all children with OI examined at the Shriners Hospital for Children in Montreal between October 1999 and October 2003. The diagnosis of OI was based on clinical findings, as described by Silene et al. All patients in whom anteroposterior radiographs of at least one hip had been performed were included in the study, and the last available x-ray was assessed for the present study. A total of 283 patients fulfilled these criteria (150 girls, 133 boys). The patients were classified according to the Silene classification, as expanded by Glorieux. The diagnostic distribution of the patients is shown in Table 1. Mean age at the time of analysis was 9.4 (range 0.3–23.3) years. One hundred seventy-eight patients had received medical therapy with intravenous pamidronate before the radiographic study. In 273 patients radiographs from both hips were available. In 10 patients only one side had undergone radiographic examination. Thus, a total of 556 hips could be assessed.

Radiologic Measurements

Figure 1 is an example of coxa vara in OI patients. All measurements were performed on standard anteroposterior radiographs of the hip (Figs. 2 and 3). The femoral neck–shaft angle was measured as the intersection of a line drawn through
the shaft of the femur with another line passing through the long axis of the femoral neck. The Hilgenreiner–epiphyseal angle was determined according to Beals’ modification of the method originally described by Weinstein et al.\textsuperscript{6,12} The normal value of the Hilgenreiner–epiphyseal angle is 16 degrees (range 0–25 degrees).\textsuperscript{6} We also measured the head–shaft angle, the angle between the longitudinal axis of the femoral shaft and a line perpendicular to the largest diameter of the femoral head.\textsuperscript{6} The lateral radiographs were also assessed in the patients with CV to evaluate femoral bowing and to identify false CV (Fig. 4). The ratio between tibia and femur length was evaluated in patients with CV in whom anteroposterior radiographs of the entire extremity (femurs and tibias on the same film) were available (n = 29). Results were compared with reference data published by Robinow.\textsuperscript{13}

Statistical Analysis

The statistical difference of the CV between the femurs that had been previously rodded and the femurs with no rodding procedure was evaluated by the chi-square test.

RESULTS

CV was present in 29 of the 283 OI patients (10.2%). The highest prevalence of CV was found in OI types III, VI, and VII, whereas no case was found among the 94 patients with OI type I (see Table 1). Fifteen patients with CV had unilateral involvement, and in 14 patients both hips were affected. Thus, 43 of the 556 evaluated hips (7.7%) were affected with CV (23 left and 20 right hips). In addition, four cases of false coxa vara were identified; these were associated with anterolateral angulation in the proximal third of the femur. On the other hand, 28 femurs had bowing in the proximal third without either false or true coxa vara. In 41 of the 43 cases of true CV, the deformity was located in the base or in the middle of the femoral neck. The trochanteric region was affected in the remaining two hips. The neck–shaft angle of the affected hips averaged 99 degrees (range 80–110 degrees) and the mean Hilgenreiner–epiphyseal angle was 68 degrees (range 40–90 degrees). The head–shaft angle averaged 104 degrees (range 78–120 degrees). Intramedullary rodding had been performed in 25 of the 29 patients (83%) and in 36 of the 43 femurs (84%) with CV. Thus, only four patients (all OI type III) with seven affected femora had no rodding procedures. In contrast, intramedullary rodding had been performed in only 140 of 513 femurs (27%) that were not affected by CV ($P = 0.007$ for the difference in prevalence of rods between femurs with and without CV). The tibia-to-femur length ratio was above the reference range in 24 of the 29 lower limbs with CV (83%) that could be evaluated. Intramedullary rodding procedures had previously been performed in 21 of these 24 femurs.

In 21 patients with 33 affected hips, we could retrieve information on the functional clinical status at the time point considered in the present study. In 24 hips (72%), flexion and external rotation were full. In 31 hips (93%), more than 90 degrees of flexion was possible. Eleven hips (33%) had no extension, and 15 hips (45%) had varying degrees of flexion contracture. Twenty-one hips (63%) had normal external rotation, and in 7 hips (21%) the range of external rotation was above normal. The average abduction in the 33 hips was 21 degrees. In 10 hips (30%), abduction was less than 10 degrees. In 9 hips (27%) it was between 11 and 20 degrees, in 11 hips (33%) between 20 and 30 degrees, and in 2 hips...
more than 30 degrees. In 17 hips (51%) internal rotation was absent or very limited. Six hips (18%) had 5 to 10 degrees of internal rotation, 2 hips (6%) 15 degrees, and one hip (3%) more than 15 degrees. Seven hips (21%) had varying degrees of external rotation contracture. Most patients with CV had a Trendelenburg gait.

**DISCUSSION**

In the present study we observed a high prevalence of CV in patients with severe forms of OI but an absence of this deformity in patients with OI type I. Thus, CV clearly was associated with the severity of bone fragility in OI. Assessment of CV in severe OI is not always simple, as the radiologic picture is often complicated by bowing of the femoral diaphysis. As previously described by the senior author (F.F.), a curvature in the proximal third of the femoral shaft can decrease the distance between the femoral head and shaft, mimicking the pattern of CV on anteroposterior radiographs, even if the neck–shaft angle is normal. In the present study we avoided this pitfall by also evaluating lateral radiographs of the femur. If CV is suspected in the presence of anterior bowing of the proximal third of the femur, a radiograph of the hip in extension should be obtained. In this manner false CV can be identified (see Fig. 4).

The pathogenesis of CV in OI is unclear. Generally speaking, CV in children could be classified as developmental, congenital, dysplastic, or traumatic. Many of these factors may have contributed to the development of CV in our patients. In developmental CV, there is an intrinsic defect in cartilage maturation and bone formation involving the proximal femoral physis and femoral neck. Its pathognomonic radiologic sign is a triangular metaphyseal fragment in the medial side of the femoral neck. Such fragments were seen in both hips of one of our patients with OI type III. In dysplastic CV, the deformity usually affects the trochanteric area; this was found in two of our patients. However, the large majority of our patients had CV secondary to deformity in the base of the femoral neck. It appears plausible that many of these cases could be classified as traumatic CV and may have been caused by malunion of a fracture or repeated subclinical fractures of the femoral neck. The majority of patients with CV had undergone femoral rodding procedures. It is possible that the biomechanical effects of femoral rods contributed to the

<table>
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<tr>
<th>OI Type</th>
<th>No. of Pts. Examined</th>
<th>No. of Patients With CV (% of all patients with same type)</th>
<th>No. of Hips Examined</th>
<th>No. of Hips With CV (% of hips of patients with same type)</th>
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<tr>
<td>I</td>
<td>94</td>
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<td>185</td>
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<tr>
<td>III</td>
<td>67</td>
<td>16 (24%)</td>
<td>131</td>
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<td>7 (8%)</td>
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<td>V</td>
<td>18</td>
<td>1 (6%)</td>
<td>36</td>
<td>1 (3%)</td>
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<tr>
<td>VI</td>
<td>10</td>
<td>4 (40%)</td>
<td>18</td>
<td>6 (33%)</td>
</tr>
<tr>
<td>VII</td>
<td>4</td>
<td>1 (25%)</td>
<td>8</td>
<td>2 (25%)</td>
</tr>
<tr>
<td>Total</td>
<td>283</td>
<td>29</td>
<td>556</td>
<td>43</td>
</tr>
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**TABLE 1. Diagnostic Distribution of the Study Population and Results of Radiologic Evaluation**

**FIGURE 2.** A, Anteroposterior x-ray of the pelvis and both hips in a child with type III OI showing severe CV with decreased femoral neck–shaft angle. B, Drawing of the femoral neck–shaft angle.
development of CV. However, the statistical association between femoral rodding and CV does not necessarily prove a causal link. Our observation may simply reflect the fact that the more severely affected OI patients are more prone to develop CV and at the same time are more likely to require rodding. Nevertheless, it may be prudent to closely evaluate such patients for the development of CV. Although CV is usually defined on the basis of the femoral neck–shaft angle, Weinstein et al suggested measuring the head–shaft angle to evaluate the varus angulation of the femoral neck. According to these authors, the head–shaft angle is more reproducible than the neck–shaft angle and more indicative of the actual deformity. We found that the mean neck–shaft angle was lower than the head–shaft angle in our patients, probably because there was no defect in the femoral neck in CV and the location of the deformity was mostly around the base of the femoral neck. The addition of the head–shaft angle to the list of measured indices therefore did not provide much clinically useful information.

As CV has been described in association with rhizomelia (shortness of the upper leg relative to the lower leg) in some cases of OI, we assessed our CV cases for rhizomelia as well. In 24 lower limb x-rays that we reviewed, we found rhizomelia in all of them. Because more than 85% of the femurs (21/24) had already been corrected surgically and rodded, and most femurs are osteotomized, this could be an interpretation for this finding. We indeed found short femurs in a considerable proportion of cases. However, the large majority of these femurs had undergone intramedullary rodding to correct bowing deformity. It therefore appears most likely

![FIGURE 3](image3.png)

**FIGURE 3.** A, Anteroposterior x-ray of the pelvis and both hips in a child with type III OI showing severe CV with increased Hilgenreiner–epiphyseal angle. B, Drawing of the Hilgenreiner–epiphyseal angle.

![FIGURE 4](image4.png)

**FIGURE 4.** A, Anteroposterior x-ray of the right hip and femur in a child with type III OI, showing false CV with decreased femoral neck–shaft angle. B, Lateral view of the hip and femur of the same child, showing that the true deformity lies in the proximal femur and not in the femoral neck. C, Anteroposterior view showing a normal neck–shaft angle of the femur of the same child, after femoral rodding and correction of bowing of the proximal femur. D, Lateral view of the femur of same child showing normal alignment of the proximal femur, after femoral rodding and correction of deformity of the proximal femur.
that short femurs were due to bony resections during rod-ding surgery rather than being caused by developmental abnormalities.

As to the clinical consequences of CV, it is well known that a decrease in the neck–shaft angle will shorten the lever arm of the abductor mechanism and thereby cause abductor insufficiency with a positive Trendelenburg sign.\(^8,18\) This was present in most of our patients. In addition, the range of motion of the hip in all of our patients was limited in abduction and internal rotation, as has also been reported in non-OI patients with CV.\(^6,11,19\)

All but the most mildly affected patients in the present cohort had received bisphosphonates at some point. In the absence of an untreated control group, we therefore cannot judge the effect of bisphosphonates on hip development. The long-term effects of bisphosphonates on hip development remain to be addressed in the future. Thus, our data cannot be used to make recommendations about medical treatment measures that could prevent such a deformity.

In conclusion, we observed a high incidence of CV in children and adolescents with severe forms of OI, particularly OI type III. We believe that the presence of CV may aggravate the functional status of these children, which is already impaired by bone fragility and multiple deformities. A high index of suspicion should be maintained in children with severe OI and regular radiologic follow-up is warranted. Thus, our data cannot be used to make recommendations about medical treatment measures that could prevent such a deformity.

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