Focal fibrocartilaginous dysplasia in the humerus

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Focal fibrocartilaginous dysplasia is an uncommon, benign bone lesion that causes deformity of the long bones in young children. It has most commonly been encountered in the proximal tibia, and very rarely in the long bones of the upper limb, that is, the proximal humerus, distal radius, ulna and proximal phalanx. Only one case of focal fibrocartilaginous dysplasia of the proximal humerus has been reported previously. The present study reports two such additional cases that were diagnosed in late childhood. The clinical presentation and radiographic findings are described with an emphasis on the natural evolution of the disease. Limb-length discrepancy is anticipated in these children in the long-term follow-up and, therefore, surgical intervention should be considered in treatment.
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Introduction

Focal fibrocartilaginous dysplasia (FFCD) is a rare, benign lesion that has generally been reported as a cause of unilateral tibia vara in children. After Bell *et al.*'s [1] first description of this characteristic lesion in three patients in 1985, additional cases involving both the lower and the upper extremity bones have been published [2–6]. Although, this benign pathologic process has been reported to occur in the proximal medial tibia previously, upper limb involvement has been described in some recent publications. Lincoln and Birch [5] were the first to describe this lesion in the proximal humerus in 1997. We report two patients with proximal humeral FFCD lesions leading to shortening and angular deformities on the affected upper limbs. The clinical presentation, radiographic appearance, and natural history of FFCD in this specific location are described.

Case reports

Case 1

An 11-year-old girl was admitted to our institution for shortening of her left upper limb, which had been noticed by her parents 2 years ago. The parents reported that the child did not have any other clinical signs, including pain or functional impairment, and did not receive any treatment during this period. The child was otherwise healthy, with no history of trauma, infection, metabolic bone disease, generalized musculoskeletal dysplasia, or neurofibromatosis. The physical examination revealed an 8 cm shortening of the left upper extremity; the left arm was significantly shorter than the right arm (Fig. 1a). No associated angular deformity was apparent. The range of motion of the left shoulder was full, except for moderate limitation in abduction (abduction = 70°). Full flexion and extension of the elbow and full pronation and supination of the forearm were observed. The neurovascular examination of both the upper extremities was normal. No soft-tissue mass, swelling, fistula tract, or erythema over the left shoulder and arm were found. The radiographic examination of the left arm showed a unilateral defect in the upper metaphyseal portion of the medial humeral cortex combined with shortening of the limb (Fig. 2a). A marked thickening of the cortex adjacent to the defect was also detected. Furthermore, an angular deformity of the humeral head resulting from varying degrees of epiphyseal closure was demonstrated. Magnetic resonance imaging (MRI) was performed to evaluate the bone lesion and to exclude an adjacent soft-tissue mass. MRI demonstrated medial physeal closure and muscular interposition in the defective area. A low signal was recorded on both T1-weighted and T2-weighted sequences in the areas corresponding to the cortical lucency on plain radiographs (Fig. 3). Areas corresponding to the sclerosis showed low signal on T1-weighted sequences and intermediate signal on T2-weighted sequences. No soft-tissue mass was found on the images.

Case 2

A 17-year-old boy with significant shortening of the left upper limb was referred to our institution for limb lengthening. The asymmetry in the upper limbs was detected by the patient and his parents about 4 years ago. The patient had a proximal humeral fracture after a fall when he was 13 years old, and this fracture has healed uneventfully by conservative treatment. The boy was healthy otherwise, with a normal antenatal and postnatal history. None of the members of the family had suffered

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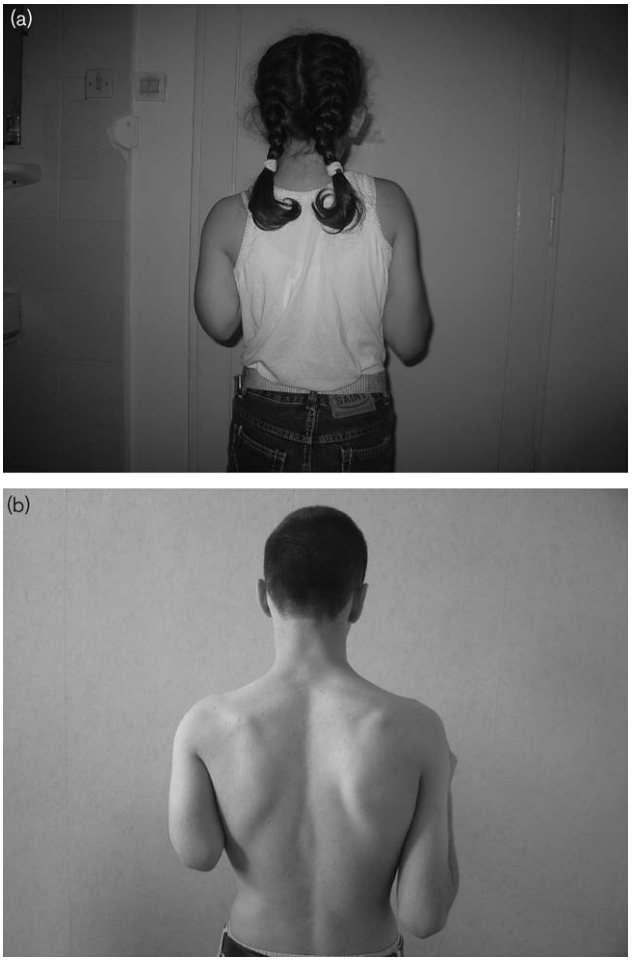
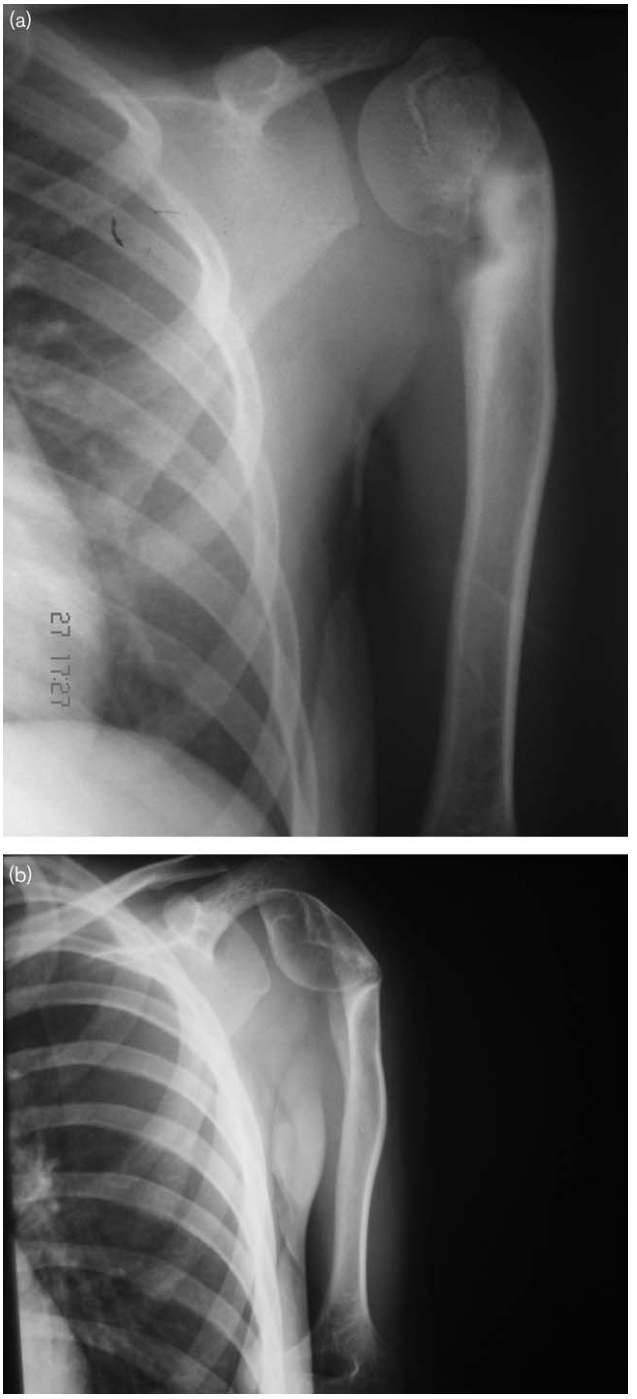
Fig. 1



Clinical picture of (a) an 11-year-old girl and (b) a 17-year-old boy with focal fibrocartilaginous dysplasia of the left proximal humerus. In both patients, there is significant shortening of the left upper limb compared with the right side. Further, also note the medial crease associated with a mild anteromedial angular deformity of the arm in the older child.

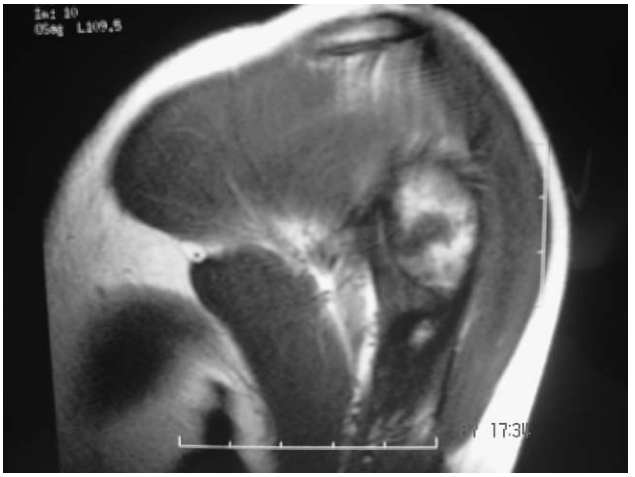
Fig. 2



The anteroposterior plain radiographs of the left arms of both patients demonstrate well defined, unilateral defects in the upper metaphyseal portions of the medial humeral cortices, combined with shortening of the limbs and marked thickening of the cortices adjacent to the defects. Furthermore, in both cases angular deformity of the humeral heads resulting from varying degrees of epiphyseal closure is demonstrated.

from a similar condition. The initial diagnosis was malunited fracture and physeal arrest. On physical examination, there was a 10 cm shortening of the left upper limb compared with the other side, with a mild unilateral anteromedial angular deformity of the arm (Fig. 1b). The range of motion of the left shoulder was limited, particularly in abduction and flexion (abduction = 45°, flexion = 45°). The elbow range of motion and forearm rotations were full. No soft-tissue mass, swelling, fistula tract, or erythema over the left shoulder and arm were observed. The neurovascular examination of both the upper extremities was normal. Radiographs showed classic features of FFCD with proximal medial radiolucency and adjacent sclerosis (Fig. 2b). MRI findings were the same as that in the other case: low signal on both T1-weighted and T2-weighted sequences in the areas corresponding to the cortical lucency on plain radiographs and low signal on T1-weighted sequences and intermediate signal on T2-weighted sequences in the sclerotic areas.

Fig. 3



On sagittal T1-weighted magnetic resonance imaging, the fibrocartilaginous defect is visualized as a hypointense region adjacent to the thickened and sclerotic cortex.

A presumptive diagnosis of FFCD was made because of the radiographic similarity of the lesion in the humerus to the characteristic appearance reported in other locations, mostly tibia. Pathological confirmation of the diagnosis was not required. The patients did not receive any surgical treatment before admission to our institution. Limb lengthening and deformity correction were planned for both patients.

Discussion

FFCD was first described by Bell *et al.* [1] in 1985, as a benign lesion that causes tibia vara. With increasing recognition of the typical radiographic appearance, more than 50 cases have been reported since then [1–6]. Although the origin of FFCD remains unknown, these reports have helped to clarify further diagnostic characteristics, pathology, and natural history of this lesion. FFCDs mostly have been reported in the proximal tibia and distal femur. Only 11 of reported cases have been in the upper limbs: in the proximal humerus, distal ulna and radius, and proximal phalanx [5,6]. Lincoln and Birch [5] reported the only lesion involving the proximal humerus in 1997. The two cases reported in this study are the additional lesions located in the proximal humerus.

A review of the literature reveals that FFCD lesions located in the lower limbs usually present in younger ages [3–5,7]. Associated deformities usually result in functional problems, such as limb-length inequalities, gait problems, and angular deformities that lead to early presentation. The patients with upper limb involvement, however, usually present at older ages [5,6]. FFCD lesions located in the upper extremity bones, such as the proximal humerus, are well masked, because the shortening and angular deformities do not result in severe functional impairment as seen in lower limb involvement.

The natural history of FFCD is still being studied, and little is known about the exact etiologic and prognostic factors. Of the tibial cases that have been observed, about 45% demonstrate spontaneous, progressive resolution [1]. Beaty and Barrett [3] reported four femoral lesions, none of them resolved because of surgically detected focal fibrous tether, whereas both the reported ulnar lesions resolved spontaneously [4,7]. The only humeral lesion reported in the literature was an early presented case [5]. In the follow-up period, the patient had two nondisplaced fractures through the area of maximal bowing after falls, with some apparent progression of his deformity. Then the patient underwent surgical treatment consisting of corrective osteotomy and internal fixation. Despite the healing of the lesion, the patient still had an 8 cm shortening of the humerus. The two patients reported in this study were late presented cases, particularly the second one who presented after skeletal maturity. Both cases had a significant shortening of the affected upper limb; the inequality and functional loss of the shoulder was more severe in the older child, suggesting an age difference in natural history. In contrast to the proximal humeral lesion reported by Lincoln and Birch, these two cases did not receive any surgical treatment intervening the natural history.

The radiographic appearance is characteristic for FFCD and adequate for the diagnosis. Plain radiographs of the affected bone demonstrate a well-defined focal area of lucency in the proximal medial cortex with sclerotic cortical thickening extending distally [3,5]. MRI and computed tomography findings confirm the cortical defect without a soft-tissue mass. MRI findings of FFCD are low signal on both T1-weighted and T2-weighted sequences in the areas corresponding to the cortical lucency on plain films and low signal on T1-weighted sequences and intermediate signal on T2-weighted sequences in the sclerotic areas. In the current patients, the radiological findings, including the plain radiographs and magnetic resonance images, were compatible with the findings described in the literature.

The differential diagnosis of FFCD should include physeal injury from infection or trauma, fracture malunion, metabolic bone disease, osseous dysplasias such as Ollier's disease and fibrous dysplasia, or a tumoral condition.

As the natural history of FFCD is still being studied and little is known about the exact etiologic and prognostic factors, treatment has been optional. Choi *et al.* [4] indicated that at least 45% of the cases of tibial FFCD

showed spontaneous resolution or progressive improvement of the deformity. The authors concluded that a proportion of cases that may not need definitive treatment may be larger than previously reported. They mentioned that many of the treated cases might have resolved on their own if they had been given the opportunity of observation without surgical correction. FFCD of the proximal humerus, however, may not be a benign condition as compared with that of the other long bones. This can be related to the location and severity of pathology. If the pathology is close to the proximal physis, growth retardation potential is highest. Both the current patients had no surgical intervention during the course of the disease, and this led to a significant shortening of the affected upper extremities.

We conclude that although FFCD of the long bones is a benign lesion, its clinical course is not uniform, particularly if it is located proximal to physis, such as the proximal humerus. When there is significant limb-length inequality in the affected limb, limb lengthening is then indicated.

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