Outcomes in Early Versus Late Presentation of Focal Fibrocartilaginous Dysplasia Affecting the Upper Extremity: A Review of 4 Cases

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Background: Focal fibrocartilaginous dysplasia (FFCD) is a rare disorder of the upper and lower extremities. In the distal ulna, a ligamentous tether emerging from the metaphysis crosses the physis and restricts growth, leading to deformity. Lesion excision before radiocapitellar subluxation has been shown to restore growth and allow remodeling. We review the outcomes of 4 patients with FFCD of the distal ulna after the occurrence of radiocapitellar incongruity.

Methods: This retrospective review examined 4 patients diagnosed with FFCD of the distal ulna from 2007 to 2015. Diagnosis was based on radiographic parameters and pathology when available. The radiographic and functional outcomes are reported.

Results: Three males and 1 female patient presented at an average of 37.5 months (range, 22 to 48 mo) with ulnar FFCD and radiocapitellar joint incongruity. The first patient had progressive radiocapitellar dislocation, poor motion, pain, and deformity. The second patient presented for a third opinion after previous surgery secondary to deformity progression and radial head dislocation. These patients required salvage procedures with creation of a 1-bone forearm. Patient 3 had frank dislocation of the radiocapitellar joint, yet maintained functional motion. This family elected for continued observation. Patient 4 had a 50% subluxation of the radiocapitellar joint and underwent tether excision and ulnar lengthening with an external fixator. Both joint congruity and deformity improved with functional forearm and elbow motion.

Conclusions: Delayed treatment of ulnar FFCD may require salvage procedures to maximize function and provide pain relief.

Key Words: FFCD, focal fibrocartilaginous dysplasia, ulna deficiency

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Focal fibrocartilaginous dysplasia (FFCD) is a rare disorder affecting the upper and lower extremities leading to limb deformity. Since the original description by Bell et al in 1985,[1] over 100 cases have been reported. FFCD more commonly affects the lower extremity in the region of the proximal tibial metaphysis, and it often presents when a child reaches ambulatory age. Other locations include the forearm, distal femur, distal tibia, humerus, phalanx, and thoracic vertebrae.[2–9] Cases affecting the upper extremity are less common, perhaps in part due to being underdiagnosed. Although the etiology remains unclear, some authors suspect a contribution from early trauma, possibly even in utero.[5,10–12] Analogous to a Madelung deformity, tethering by the fibrous tissue across the physis may focally restrict growth as per the Hueter-Volkmann Law.[2,13–15] In Bell original article, 2 of the 3 patients that underwent surgery were found to have tissue that resembled cartilage at the location of the deformity. This finding has been supported by numerous case reports that followed.[3,16] Management often includes observation, while corrective osteotomies are only required in the lower extremity in cases of severe deformity.[7,17,18] Despite reports of good outcomes with observation for the proximal tibia, the outcomes with nonoperative management in the upper extremity may not be as successful. Smith et al[8] described 7 cases of FFCD affecting the upper extremity, with 5 of the cases involving the forearm. The spectrum of results varied from small differences in limb length, to pain, loss of motion, and frank dislocations of the radiocapitellar joint. We report 4 new cases of FFCD in the upper extremity to emphasize the consequence of late recognition. We also review the current literature on this topic, and summarize the outcome differences between upper and lower extremity cases.

METHODS

During 2007 to 2015, we treated 4 cases of FFCD of the upper extremity at 2 of our institutions (Shriners Hospital for Children, Philadelphia, PA; Shriners Hospital for Children, Greenville, SC). This study was approved by our institutional review board. The hospital database was used to access patient charts and radiographs. All 4 cases affected the forearm at the ulna and radius. Diagnostic criteria included unilateral angular deformity in a long bone with a characteristic radiolucent lesion, surrounding

sclerosis, and a cortical defect. Three male patients and 1 female patient were identified. Ages at time of presentation to our institution were 22 months to 4 years (average 37.5 mo).

Case Summaries

Case 1

A healthy 22-month-old male was referred to our office for a bump on the right forearm noticed 6 months before the office visit. Examination demonstrated a radial head that was able to be subluxed with provocative maneuvers and a shortened ulna. Forearm range of motion measured 80 degrees of supination and 30 degrees of pronation. Radiographs (Fig. 1) demonstrated deficiency of the distal ulna, a lucent lesion in the distal diaphysis, and surrounding sclerosis. This was consistent with FFCD. The radius was bowed with subluxation of the radiocapitellar joint.

After initial observation, the patient returned at 2 years and 7 months of age with pronation limited to neutral, a prominent radial head, and an obvious forearm bow. Radiographs showed a dislocated radial head and a shortened ulna. Again, observation was chosen. At age 7, the patient complained of lateral elbow pain. Examination showed a short forearm with bowing that was fixed in complete supination. Radiographs demonstrated a dislocated radial head, a short ulna, and the persistent FFCD lesion (Fig. 2). Owing to the fixed supination and pain, the family opted for surgery. An osteotomy of the radius and ulna was performed with creation of a 1-bone forearm, resection of the proximal radius, and excision and curettage of the FFCD lesion. Fixation of the 1-bone forearm in 25 degrees of pronation was performed with a 2.7 mm dynamic compression plate. Pathology demonstrated benign bone with a focus of dense fibrous tissue. Six weeks postoperatively, the patient was neurologically intact and pronation of the forearm was maintained at 25 degrees. Radiographs demonstrated callus formation at the osteotomy between the radius and ulna (Fig. 3). At most recent follow-up at age 9, the patient had no complaints, reported participation in sports without pain, and maintained full elbow flexion and extension. The forearm position was fixed in slight pronation. Ultimately, the selected treatment offered a more functional position of the forearm and eliminated the patient's pain.

Case 2

A healthy 4-year-old male was brought to our office for a third opinion regarding his left forearm deformity. Two years prior, the patient was diagnosed with FFCD of the left ulna, and underwent excision of the lesion at an outside hospital. Since then, the deformity of the forearm progressed and a bump on the lateral aspect of the elbow formed. The patient denied pain in the elbow or forearm. Range of motion demonstrated 80 degrees of supination and 20 degrees of pronation. The radial head was clinically dislocated with a shortened and deformed forearm. The ulnar aspect of the forearm demonstrated a healed but hypertrophic scar from previous surgery. Neurovascular

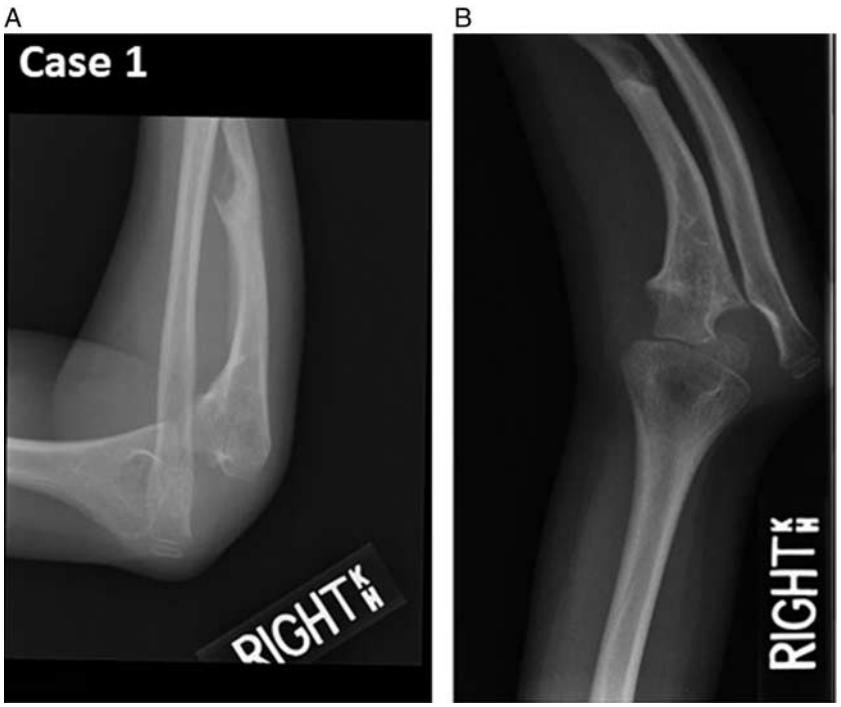

FIGURE 1. A and B, Case 1: seen above is the right forearm of a 22-month-old male with typical radiographic findings consistent with focal fibrocartilaginous dysplasia: deficiency of the distal ulna, a lucent lesion in the distal one third of the diaphysis, and surrounding sclerosis. The radiocapitellar joint is subluxed.

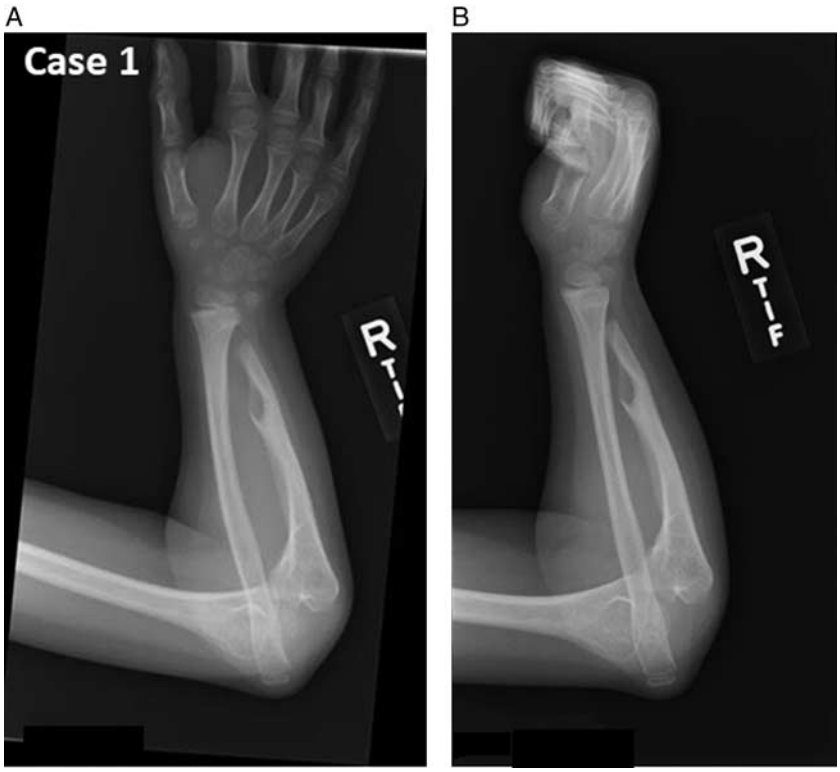

FIGURE 2. A and B, Case 1: radiographs at age 7 years demonstrate the dislocated radiocapitellar joint and shortened ulna.

examination was normal in the affected extremity. Radiographs showed a shortened ulna with residual lucency in the distal ulna and surrounding sclerosis consistent with FFCD. The radius was bowed and the radial head dislocated (Fig. 4). The family wished to proceed with a 1-bone forearm surgery to improve the forearm position and deformity. In January 2013 at age 4, the patient had an excision of the proximal radius, osteotomies of the radius and ulna, and creation of a 1-bone forearm using a 7 hole, 2.7 mm dynamic compression plate. Postoperative follow-up demonstrated appropriate positioning of the forearm in 20 degrees of pronation and a neurovascular intact upper extremity. Radiographs obtained at 4 months postoperatively demonstrate complete union of the 1-bone forearm procedure (Fig. 5). Two-year follow-up at age 7, the patient had no complaints and demonstrated forearm position fixed at 30 degrees pronation, full elbow flexion and extension, and a normal neurovascular exam. The selected treatment allowed for removal of the prominent radial head, created a stable forearm, and positioned the forearm in a functional position for daily activities.

Case 3

A healthy 32-month-old male was brought to our clinic for a progressive left forearm deformity that began at age 8 months. Physical examination demonstrated full elbow flexion and extension. Forearm pronation and supination was 50 and 50 degrees, respectively. Palpation demonstrated a dislocated radial head posterior and lateral. Neurovascular examination was normal in the affected extremity. Radiographic evaluation showed a posterior and laterally dislocated radial head, bowing of the ulna, and an area of cortical lucency with surrounding sclerosis suggestive of FFCD of the ulnar shaft. The family opted for observation at this time. Over the next 2 years, the patient had no change in his physical examination or radiographs. At the latest follow-up at age 11, he continued to be pain-free and with full elbow flexion and extension despite the radial head dislocation. Supination and pronation measured, 50 and 80 degrees, respectively. Given his function and lack of complaints, continued observation was elected.

Case 4

A healthy 4-year-old female presented to our clinic for right forearm deformity. She had full motion of the elbow and wrist, with slight loss of active forearm pronation. Initial radiographs showed bowing of the forearm, and an area of radiolucency in the distal ulna with surrounding sclerosis, consistent with FFCD (Fig. 6). Observation was selected at this time. Two years later, she returned with regular pain at the elbow, but maintained full forearm motion. Radiographs showed no changes from prior imaging. Repeat evaluations were unchanged and an magnetic resonance imaging (MRI) was performed that demonstrated a 12 mm lesion in the area of the distal ulna. At age 10 years, she returned with no new complaints or limitations, but there was a prominence at the lateral elbow. She had a 30-degree varus carrying angle, a prominent radial head, full elbow flexion and extension, and a mild decrease in pronation. Radiographs showed a 50%

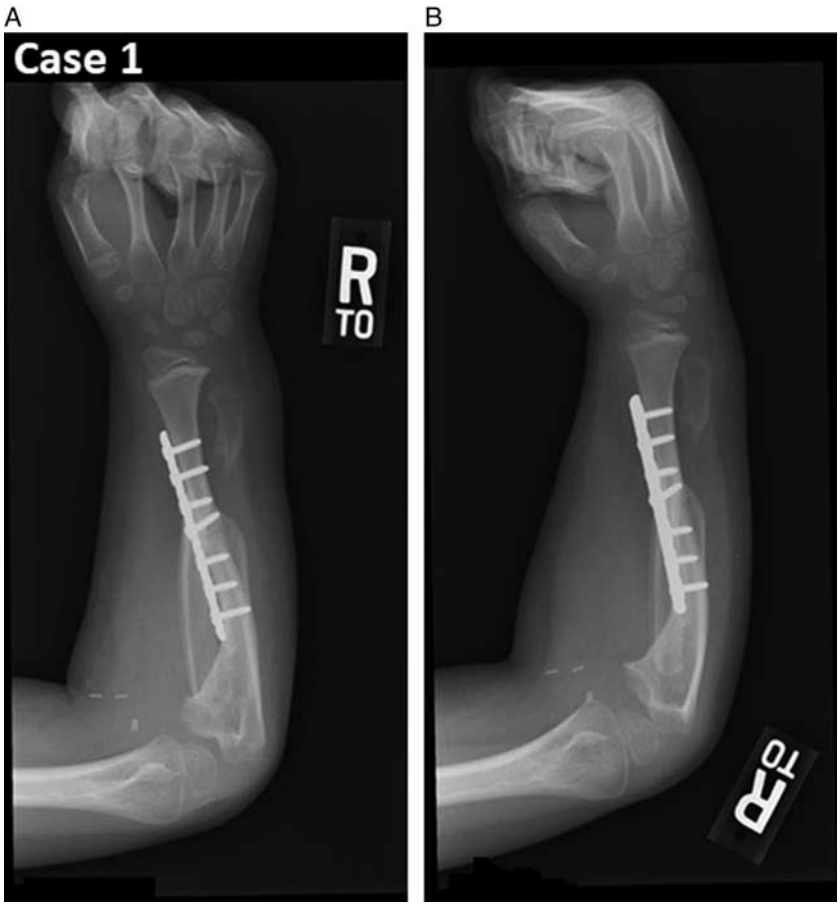

FIGURE 3. A and B, Case 1: radiographs at 6 weeks postoperative demonstrate the right forearm after resection of the proximal radius, excision of the ulnar focal fibrocartilaginous dysplasia lesion, osteotomy of the radius and ulna, and creation of the 1-bone forearm with a 2.7 mm compression plate. Callus formation is seen at the osteotomy site.

subluxated radial head, and a forearm with a 45-degree bow (Fig. 7). Surgical intervention was elected in order to address the pain, limited pronation, and newfound radiocapitellar subluxation. In December 2014 at age 10, she underwent an ulnar closing wedge osteotomy, application of an external fixator to restore ulnar length, and removal of the FFCD lesion (Fig. 8). Ulnar lengthening with the external fixator over time demonstrated improvement in her deformity. At follow-up a year later, she demonstrated a congruent radiocapitellar joint (Fig. 9), full supination and pronation, and elbow motion from 0 to 125 degrees. The selected treatment allowed for correction of joint congruency, return to normal range of motion, and a pain-free extremity.

DISCUSSION

Since its first report in the literature in 1985, numerous cases have been described affecting the upper limbs, lower limbs, and even the axial skeleton. The etiology is yet to be clarified, but the histologic presence of either dense fibrous tissue, or combinations of fibrocartilage and hyaline cartilage can be seen.[15] Jouve et al[19] coined the term "fibrous periosteal inclusion" and hypothesized that the deformity was caused by a soft tissue anchor on the periosteum preventing its sliding during longitudinal bone growth. Kazuki et al[20] described 5 cases of ulnar dysplasia similar to FFCD, but preferred the term "focal ulnar cortical indentation" to emphasize the histologic difference in their patients, which demonstrated tissue resembling periosteum and not fibrocartilage. They believed that FFCD lesions attached to the ulnar metaphysis, whereas focal ulnar cortical indentation reached the epiphysis. However, we feel that these 2 entities are likely the same pathologic process on a single disease spectrum. Some lesions contain more acellular fibrous tissue, and some may contain more elements of fibrocartilage. We agree with Kim et al[15] who argue that the histopathology of FFCD is variable and the presence of fibrocartilage is not an essential feature of the FFCD lesion.

Radiographic appearance is rather uniform in all cases of FFCD. Radiographs demonstrate an angular deformity, a radiolucent cortical break, and presence of sclerosis surrounding the lesion. This appearance has become classic and advanced imaging, such as MRI, is not routinely recommended to make the diagnosis. MRI findings are well characterized by hypointensity on both

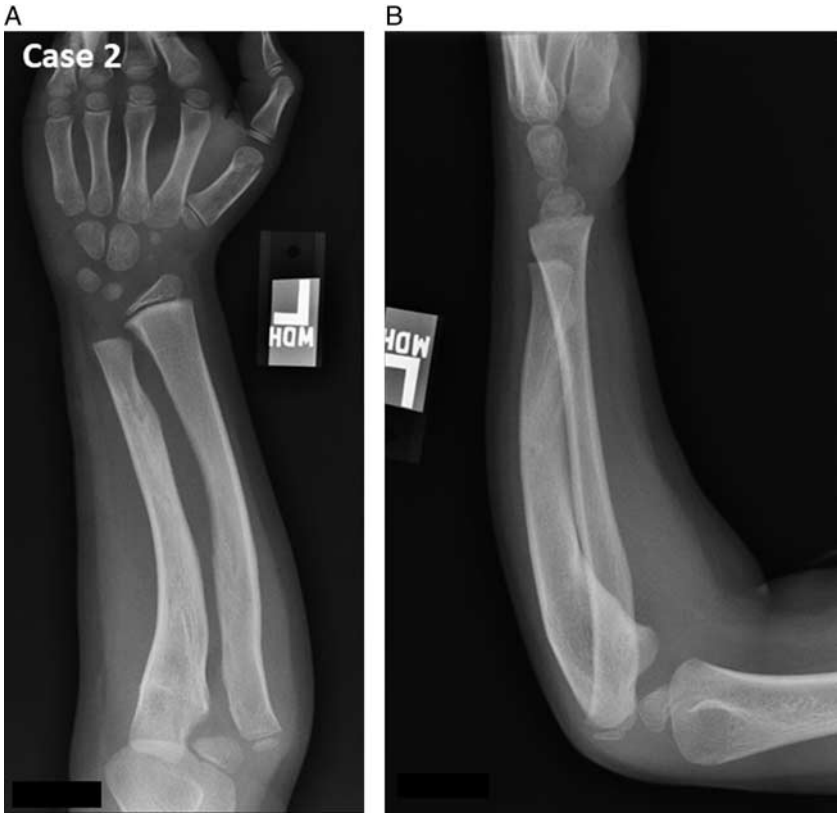

FIGURE 4. A and B, Case 2: radiographs of the left forearm in a 4-year-old male demonstrating a focal fibrocartilaginous dysplasia lesion in the distal ulna, a shortened ulna, and dislocated radial head.

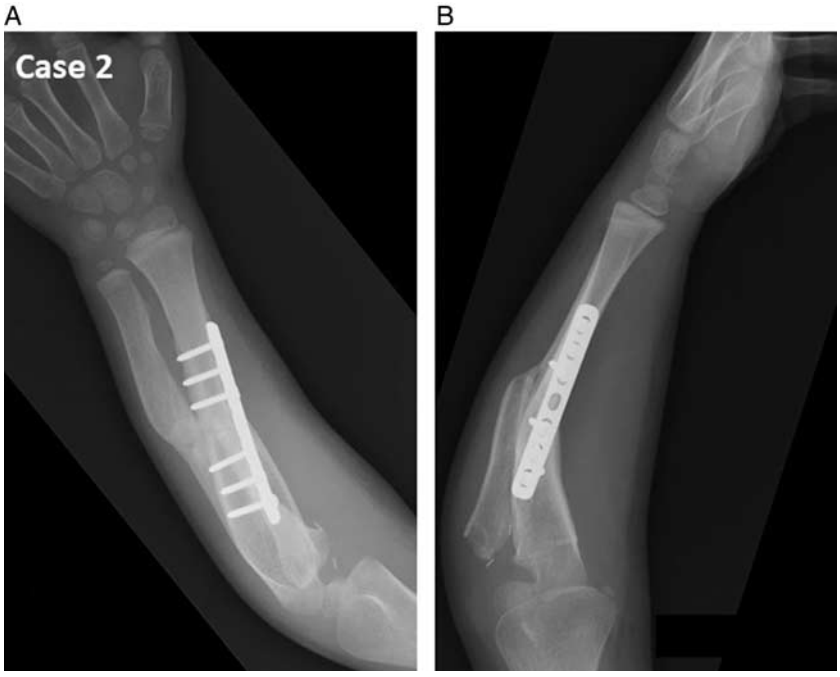

FIGURE 5. A and B, Case 2: postoperative follow-up at 4 months demonstrates union at the osteotomy site.

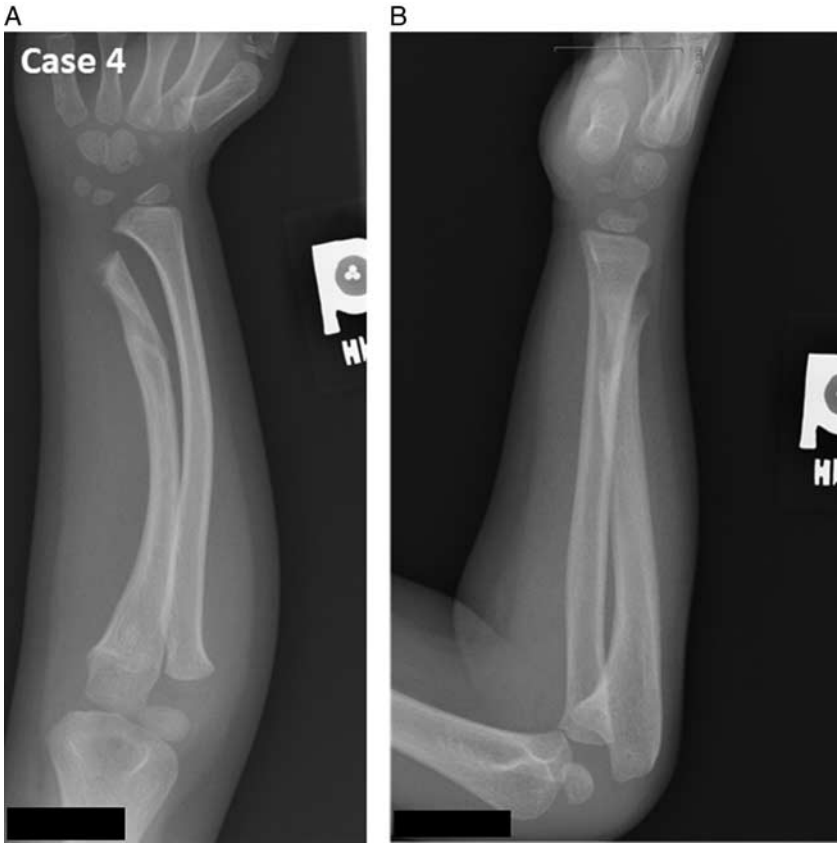

FIGURE 6. A and B, Case 4: radiographs of the right forearm in a 4-year-old female with bowing and evidence of the focal fibrocartilaginous dysplasia lesion in the distal ulna. The radiocapitellar joint is congruent.

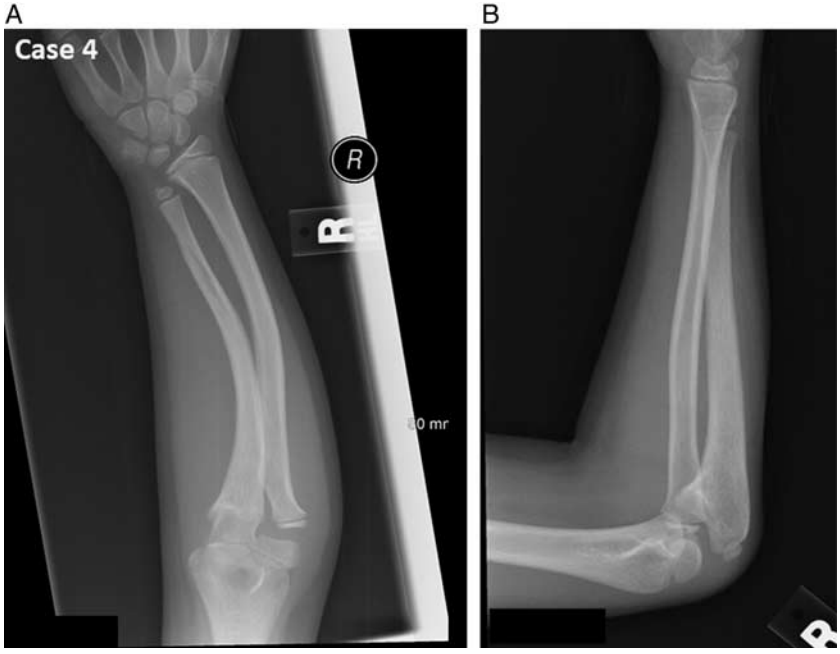

FIGURE 7. A and B, Case 4: radiographs at age 10 demonstrate the deformity progression of the right forearm with 50% subluxation of the radiocapitellar joint.

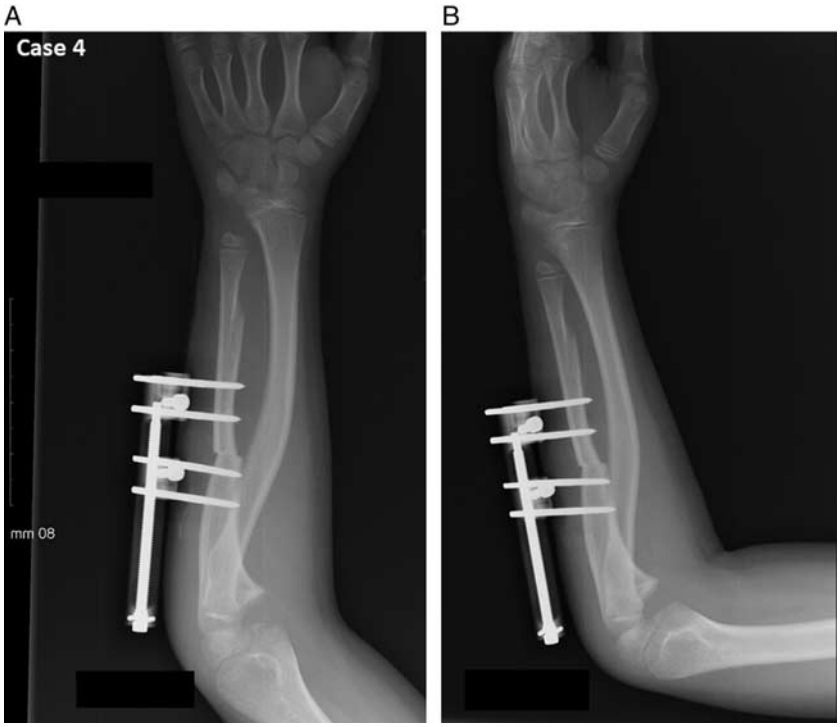

FIGURE 8. A and B, Case 4: postoperative radiographs demonstrating excision of the focal fibrocartilaginous dysplasia lesion, ulnar osteotomy, application of uniplanar external fixator.

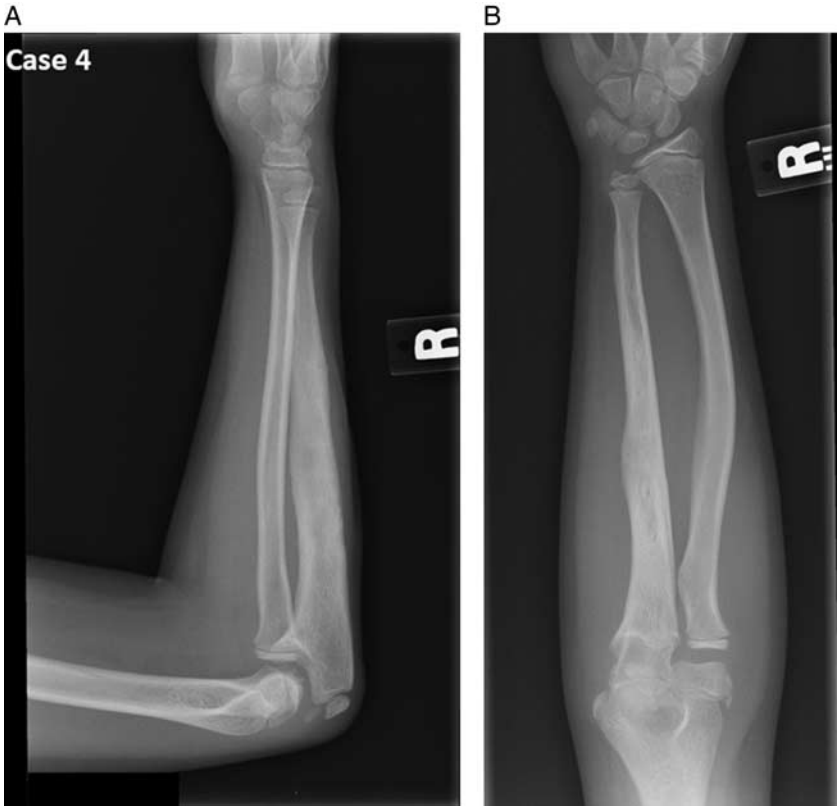

FIGURE 9. A and B, Case 4: radiographs at age 11 after removal of the external fixator demonstrating a well-healed osteotomy site and a congruent radiocapitellar joint.

T1 and T2 sequences with lack of enhancement on gadolinium administration.[16,21,22] Ringe et al[23] included an ultrasonography evaluation of an FFCD lesion causing unilateral tibia vara in a 14-month-old male. The imaging revealed a hypoechoic area visible upon the cortex of the meta-diaphysis. Computed tomography does not yield much additional information beyond demonstrating the cortical defect and marginal sclerosis, which is apparent on plain radiographs. A 99m-technetium bone scan is also of little value. One series by Albinana et al[2] demonstrate slight uptake in the affected area in 3 cases causing tibia vara, whereas Jouve et al[24] did not find any abnormality.

When examining the reports of FFCD affecting the upper extremity, the ulna is affected in the majority of cases, followed by the radius, humerus, and the phalanx. A summary of the published upper extremity case reports is found in the Appendix, Supplemental Digital Content 1, http://links.lww.com/BPO/A160. Patients with FFCD of the upper extremity have worse outcomes with conservative management. The first cases were reported by Lincoln and colleagues in 1997. One patient had an ulnar deformity and 1 patient had a proximal humerus deformity. The ulnar deformity resolved after 2 years of observation and resulted in <1 cm of length discrepancy. However, the humerus deformity progressed and the patient sustained 2 fractures through the FFCD lesion on 2 separate occasions. The deformity required an osteotomy and internal fixation. Smith et al[8] reported on 7 FFCD cases affecting the upper extremity. In their series, the lesion was found within the radius in 2 cases, within the ulna in 3 cases, and within the phalanges in 2 cases. Excision of the FFCD lesion, ulnar osteotomy, and lengthening with an Ilizarov frame was required in 1 case affecting the ulna due to progressive deformity and radiocapitellar joint dislocation. The 2 lesions involving the radius improved with conservative management with acceptable results.

Kazuki and colleagues reported 5 cases of FFCD affecting the ulna that required surgical intervention due to deformity and radiocapitellar joint dislocation. Case 1 required multiple unsuccessful procedures and persistence of the deformity. In their second and third case, a 1-bone forearm procedure was performed as a salvage operation. The remaining 2 cases were successfully treated after osteotomies and lengthening with external fixation.

Gottschalk et al[25] reported on 3 additional cases affecting the ulna and radiocapitellar joint congruity. In 2 of their cases, the FFCD lesion and soft tissue tether were excised before radiocapitellar dislocation. A progressive correction of the deformity was achieved over time with improvement in the radiocapitellar alignment. In their third case, excision of the FFCD tether and an osteotomy at the ulnar was performed with fixation over a rush rod. Over 5 years, the deformity improved and the subluxed radial head relocated to an improved position. However, long-term data are not available. Verhoeven and De Smet[26] reported a case affecting the ulna in a 26-month-old female. Surgery was indicated for a progressive deformity after a trial of conservative management. At time of surgery, the FFCD tether was excised and histopathology demonstrated fibrous tissue with focal areas of metaplastic bone. Unfortunately, the authors did not report on the outcome of this tether release. Nakura and colleagues recently described ulnar FFCD in a 33-month-old male with progressive deformity and radial head dislocation. Treatment consisted of lesion excision, osteotomy, and lengthening of both radius and ulna with gradual reduction of the radial head. At 2 years follow-up, the child had a successful outcome with deformity correction, but was limited to 30 degrees of forearm pronation.[27]

Our study does have several limitations. One limitation is that only case number 1 has pathologic confirmation, and we do not have available histology slides. However, the operative reports confirmed the presence of fibrous-like tissue at the surgical site consistent with the radiographic findings. In addition, the radiographic and clinical findings are diagnostic, and we are not aware of any other diagnoses that mimic this condition. Another limitation is the small number of patients from which to draw conclusions. However, we attempt to draw some conclusions by examining the literature and applying it to our patient series. Upper extremity FFCD rarely self-corrects with observation. Late recognition leaves only salvage operations. Lesion excision before radiocapitellar dislocation maybe all that is needed to return the forearm to normal growth. At the very least, close follow-up is encouraged with the push for surgery as soon as radial head subluxation is noticed. Our report adds 4 additional cases to the literature, which may help future clinicians recognize this problem and intervene appropriately. Another limitation is the short follow-up as none of the patients reached skeletal maturity. Thus, drawing long-term conclusions on patient outcomes is impossible. Ultimately, a follow-up publication is needed at skeletal maturity. Finally, our data do not include any validated patient outcome questionnaires, which would be beneficial in any future publications.

Our series included 4 patients with FFCD. Unfortunately, none of our patients were treated early enough to have excision of the FFCD lesion alone. These 4 patients were observed for extended periods of time and 2 required salvage procedures, 1 required an ulnar osteotomy and lengthening, and the fourth developed a shortened forearm with gross deformity. The FFCD lesion of the ulna is not likely to spontaneously resolve as in the tibia. We believe that early recognition and excision of the lesion is paramount to preventing progressive deformity, radiocapitellar dislocation, pain, and limitations in function. There is possibility that early excision may allow for resumed normal growth. If a child presents late, or the diagnosis is missed, a salvage procedure may be the only remaining option.

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