Timing of Forearm Deformity Correction in a Child With Multiple Hereditary Exostosis

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Abstract

Multiple hereditary exostosis (MHE) can cause progressive skeletal deformities (eg, ulnar shortening, bowing, radial head dislocation) in the upper extremity. Various procedures have been used to correct forearm deformity in children with MHE. The timing of these procedures, however, is controversial.

An 11-year-old boy with known MHE presented 7 months after onset of left elbow pain and loss of flexion-extension to 40° to 120°. Radiographs showed a distal ulna osteochondroma, ulnar shortening, bowing, and radial head dislocation. Eighteen months after ulnar osteotomy, radial head reduction, and external fixation, he had motion from 0° to 135°.

This case demonstrates that later stage MHE-induced forearm deformities can be successfully surgically corrected. Given possible recurrence during growth, there may be an indication for waiting until the patient is closer to skeletal maturity before proceeding with corrective forearm procedures.

Multiple hereditary exostosis (MHE) is an autosomal-dominant genetic disorder characterized by development of several osteochondromas. These tumors typically appear at the metaphyseal ends of long bones and may cause progressive skeletal deformity as they grow. MHE specifically affecting the forearm often consists of ulnar shortening, leading to forearm deformity and eventual radial head dislocation. Surgical correction can entail ulnar osteotomy and lengthening, but studies in the literature are unclear as to the timing of such a corrective procedure.

We report the case of a patient with MHE who underwent operative forearm deformity correction and reduction of a dislocated radial head several months after the onset of pain and range-of-motion limitations. The patient's functional outcomes were excellent. His case suggests that corrective surgery can be successful even when performed at a later stage of deformity. The patient and his parents provided written informed consent for print and electronic publication of this case report.

Case Report

An 11-year-old boy, with a medical history significant for MHE diagnosed at age 4, was presented to the outpatient pediatric orthopedic surgery clinic with several months of left elbow pain. The boy stated he first noticed the pain, an aching sensation throughout the elbow without radiation, 6.5 months earlier and denied any history of trauma. The pain intensified and became associated with progressive loss of left elbow extension. The patient denied any neurologic deficits. Of note, he was the second of 2 children, born by normal spontaneous vaginal delivery, with normal developmental milestones. His sibling did not have MHE or similar symptoms. He had no prior surgeries, was not taking any medications, and had no known drug allergies.

Physical examination revealed a prominence and tenderness over the left radial head. In addition, there was an obvious varus deformity of the left forearm with ulnar deviation of the wrist. Left elbow range of motion (ROM) was restricted from 40° to 120° of flexion-extension and nearly full supination and pronation. The contralateral (right) elbow had ROM from 0° to 130° of flexion-extension with mildly restricted supination and pronation. The functioning of the extensor pollicis longus, flexor pollicis longus, and interosseous muscles were intact. There were no other areas of pain or deformity in the upper extremities.

Anteroposterior and lateral radiographs of the left forearm and clinical photographs were obtained at the first visit (**Figures 1A-1D**). A distal ulnar exostosis was visualized projecting over the region of the interosseous membrane on both views. There were associated ulnar foreshortening and mild apex dorsal ulnar bowing. In addition, posterolateral radial head subluxation and near dislocation were seen. Given the natural history of MHE and the position of the distal ulnar exostosis, it was presumed that this lesion contributed to the radiocapitellar subluxation/near dislocation. Magnetic resonance imaging of the left elbow without contrast (**Figures 2A-2C**), obtained 9 days later, showed posterolateral radial head dislocation with entrapment of the annular ligament within the radiocapitellar joint space. Osteochondral impaction injuries of the anterior radial head and capitellum and injuries of the anterior trochlea and coronoid process were seen.

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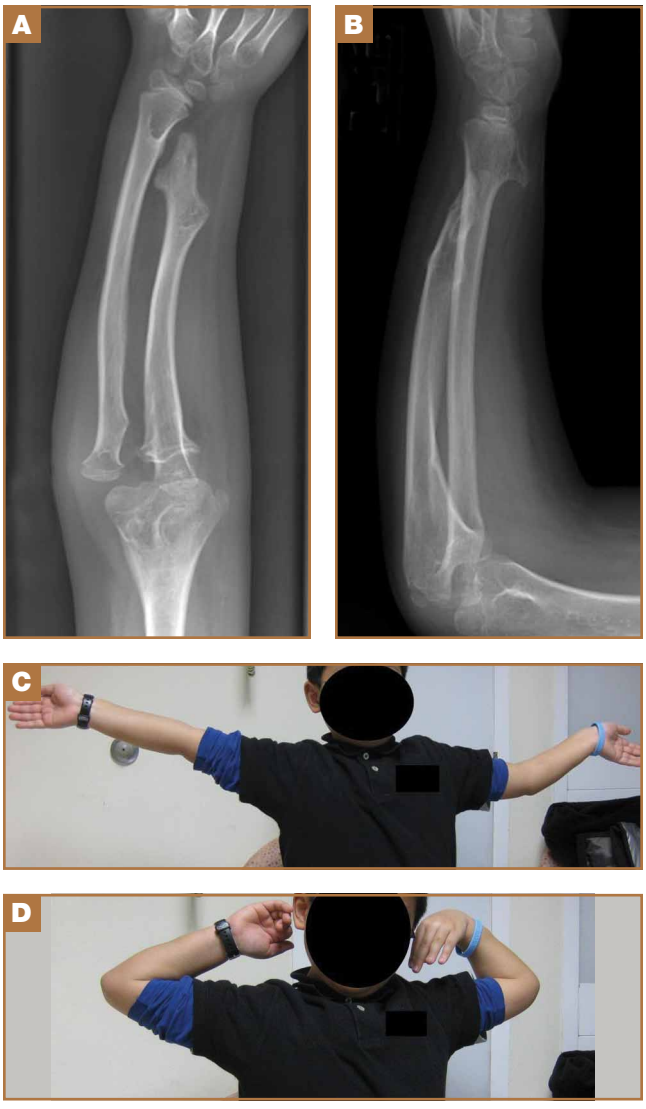

Figure 1. Radiographs and clinical photographs of patient's left forearm at initial presentation. (A) Anteroposterior and (B) lateral radiographs show distal ulnar exostosis with associated ulnar shortening and ulnar bowing. Posterolateral radial head subluxation and near dislocation are also apparent. (C,D) The patient's elbow range of motion was limited to 40° to 120°.

Given the progressive left elbow pain, restricted ROM, and forearm deformity with ulnar shortening and radial head subluxation/dislocation, corrective surgery was indicated. Seven months after pain onset, the patient was taken to the operating room, and a multiplanar external fixator was placed. Specifically, a 135 two-thirds ring was placed orthogonally proximally along the olecranon, and a distal ring was applied; these rings were fixed in position with pins and wires. An ulnar osteotomy was subsequently performed under direct visualization with reduction of the angular deformity of the ulna. This resulted in reduction of the left radial head. The ulnar exostosis was noted to not be impinging. Intraoperative ROM assessment revealed full extension and flexion. The patient tolerated the procedure without complication and was started on a 2-week postoperative course of cephalexin for pin-tract infection prophylaxis.

The patient progressed well after the operation. At his first postoperative visit, 9 days after surgery, pain was well-controlled, and left elbow ROM was from 15° to 90° of flexion-extension. Further lengthening was performed to correct the ulnar shortening until the distal radioulnar joint appeared well-balanced on dedicated wrist films. By 5 weeks after surgery, ROM had improved to 0° to 100°, and radiographs showed interval callus formation. By 3 months after surgery, ROM had improved to 0° to 135°. Radiographs and clinical photographs from this time are shown in **Figures 3A-3D**.

Five months after surgery, the external fixator was removed, and a temporary ulnar gutter splint was placed. Radiographs from the latest, 18-month follow-up (**Figures 4A, 4B**) showed maintained anatomical alignment and reduction of the radial head. The patient was now 13 years old, and elbow ROM was from 0° to 135° with full supination and pronation. He was returning for outpatient follow-up every 6 months, with forearm radiographs taken until skeletal maturity.

Discussion

MHE is an autosomal-dominant disorder with nearly complete penetrance.[1] Implicated in the development of MHE are mutations in the exostosin genes—namely, *EXT1* (on chromosome 8q23-q24) and *EXT2* (on chromosome 11p11-p13), both of which have tumor suppressor properties, and, in rare cases, *EXT3* (on chromosome 19p).[2] These mutations lead to aberrant regulation of cell surface heparin sulfate proteoglycans and consequently of cartilage hyperproliferation. The true prevalence of these mutations is not precisely known, as many patients may be asymptomatic and undiagnosed. Studies have estimated that 0.9 to 2 in 100,000 Caucasians have MHE, with other populations in Guam and native communities in Canada experiencing prevalences as high as 100 to more than 1000 in 100,000 people.[1] The osteochondromas, which typically grow at a rate tantamount to the general growth of the patient while the physes are open, are often present on the distal ends of rapidly growing long bones.

MHE has a variety of anatomical sequelae. Short stature is common, and approximately 40% of patients have a limb deformity, 10% have limb-length inequality, and 8% have limb malalignment.[1] In a prognostic study, Alvarez and colleagues[2] evaluated the anatomical burden in 79 patients with MHE and concluded that patients with the *EXT1* mutation were shorter than patients with the *EXT2* or *EXT3* mutation and had more lesions. With respect to upper extremity involvement, MHE often leads to ulnar shortening relative to the radius because the ulnar physis undergoes less longitudinal growth and has a smaller cross-sectional area. Radial bowing, also often present, leads to ulnar wrist deviation. Consequently, radial head subluxation or dislocation is found in 22% to 33% of patients with MHE, with redislocation rates reported to range from 25% to 50% by 2 years after initial reduction.[3-5] Given that the degree of forearm involvement correlates with overall sever-

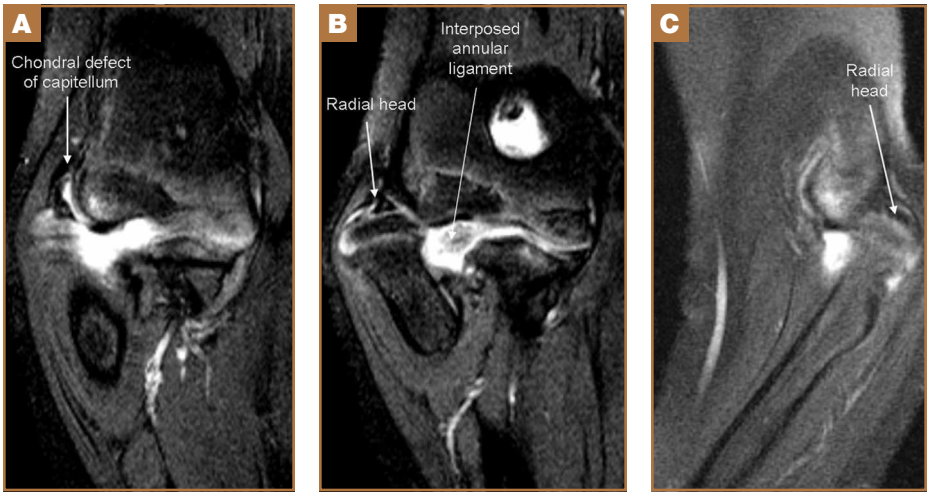

Figure 2. At initial presentation, (A,B) coronal and (C) sagittal magnetic resonance imaging of left elbow shows posterolateral radial head dislocation with interposition of annular ligament.

ity of MHE, Taniguchi[6] developed a 3-group classification system for MHE. Group 1 has no involvement of the distal forearm, group 2 has involvement of the distal radius or ulna without shortening, and group 3 has involvement of either the distal radius or ulna with shortening. Our patient in this report presented with a Taniguchi group 3 deformity.

Surgical correction of forearm deformities in the setting of MHE is controversial but may be indicated for persistent pain, loss of ROM, progressive deformities, symptomatic radial head instability, worsening cosmesis, or decreased proportional ulnar length.[7-9] Treatment modalities vary. In a prospective analysis of 14 forearms, Ishikawa and colleagues[10] showed that simple excision of osteochondromas is effective in patients with an isolated tumor of the distal ulna but is less effective when present in both the distal radius and ulna. The Sauvé-Kapandji procedure, which involves distal radioulnar joint fusion coupled with the intentional production of a distal ulnar pseudarthrosis, can improve forearm rotation.

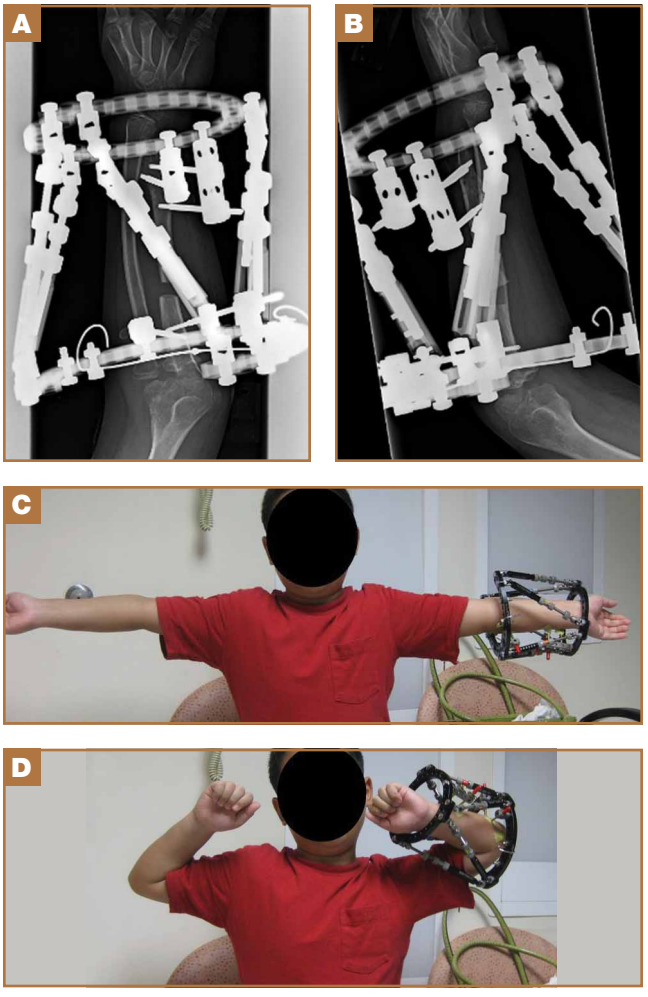

Figure 3. Radiographs and clinical photographs of left forearm after index procedure. (A) Anteroposterior and (B) lateral radiographs show placement of external fixator with ulnar diaphyseal osteotomy. (C,D) The patient's elbow range of motion improved to 0° to 135°.

Figure 4. At 18-month follow-up, (A) anteroposterior and (B) lateral radiographs of left forearm show maintained anatomical alignment, reversal of ulnar shortening, and reduction of radial head.

When combined with simple excision, this operation can also improve wrist stability and radiographic appearance.

Ulnar lengthening procedures can also be performed to restore carpal balance, which was achieved in our patient.[1] Ulnar osteotomy and gradual lengthening with application of an external fixator can be used to reduce a radial head dislocation through traction.[11] The timing of this correction, however, is controversial. Many believe early intervention maximizes remodeling potential and can be performed prophylactically before the radial head dislocates. Specifically, Ip and colleagues[12] reported a high degree of satisfaction, as well as good forearm ROM and restored radial articular angles with improved radiographic outcomes, in a small series of MHE patients treated at the mean age of 7.6 years for complex forearm deformities. Those authors consequently advocated for early and aggressive treatment. Similarly, Vogt and colleagues[5] noted improved ROM and wrist/forearm radiographic parameters in their retrospective series of 12 children (mean age, < 10 years) treated for MHE-related ulnar shortening by distraction osteogenesis. Despite observing mild recurrences of radial malformation and ulnar shortening, the authors recommended performing surgery early in the disease process. Nonetheless, the disadvantage of early intervention is that the child's continuing growth can lead to recurrent deformity and the need for multiple, invasive procedures. Given the burden of MHE on other limbs, this may heavily impact the amount of time a child spends in the hospital during his or her formative years.

Others advocate operating at later stages to minimize the chance for repeat procedures. Abe and colleagues,[13] reporting on a series of 10 children, stated that recurrences can and should be avoided by delaying lengthening procedures until after age 10. Moreover, both Arms and colleagues[9] and Stanton and Hansen[14] proposed postponing reconstructive surgery, as their retrospective analyses of more than 100 patients combined demonstrated good functional outcomes with delayed intervention. Little is known, however, about the adverse effects of postponing surgery.[5] There may be increased risk for arthritis and increased technical difficulty in restoring and maintaining radiocapitellar alignment. In addition, there are no clear guidelines as to the upper limit of treatable MHE deformity.

Conclusion

We have reported a case of ulnar deformity correction and radial head reduction performed 7 months after progressive radial head dislocation and loss of elbow motion. This case showed that ulnar osteotomy, lengthening, and closed reduction can be successful even at an advanced stage of deformity. Good outcomes may be achievable even when corrective surgery is performed later, closer to skeletal maturity, which we recommend. Larger prospective studies are needed to confirm these findings in the broader population of patients with MHE.

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This paper will be judged for the Resident Writer's Award.