One-bone forearm procedure for Hajdu–Cheney syndrome: a case report

Abdo Bachoura · Sidney M. Jacoby · A. Lee Osterman

Published online: 10 July 2013
© American Association for Hand Surgery 2013

Abstract One-bone forearm surgery is generally regarded as one of the last available salvage procedures that could be used to treat patients with longitudinal forearm instability secondary to a congenital, oncologic, or a post-traumatic etiology. We performed this procedure on a 23-year-old patient with longitudinal forearm instability secondary to Hajdu–Cheney syndrome, which is a rare genetic disorder characterized by generalized ligamentous laxity, skeletal dysplasia, acro-osteolysis, and generalized osteoporosis. The patient developed shoulder pain secondary to overuse 28 months following treatment, and was managed conservatively. Eight years after surgery, the patient had not undergone any additional procedures, had no pain, reported a Quick Disabilities of the Arm Shoulder and Hand score of 21, and was completely satisfied with treatment. Although OBF procedure is a radical first-line salvage option, in unique circumstances and appropriate patient selection, it may provide acceptable, durable, and predictable results.

Introduction

Hajdu–Cheney syndrome is a rare genetic disorder characterized by generalized skeletal dysplasia, ligamentous laxity, osteoporosis, and acro-osteolysis of the fingers and toes. Additional musculoskeletal manifestations include cervical spine instability, kyphoscoliosis, serpentine fibula, and short stature [2, 4, 6, 10, 14]. Mutations located within exon 34 of the NOTCH 2 gene have been implicated in the cause of this disease [13]. The NOTCH2 gene mutation leads to a deficient Notch 2 receptor protein. The Notch receptor family plays a

regulatory role in skeletal development and has been found to maintain bone marrow progenitors by suppressing osteoblast differentiation [3]. This report describes the utility of the one-bone forearm procedure for the treatment of a patient with forearm instability secondary to Hajdu–Cheney Syndrome.

Case report

In May of 2005, a 23-year-old right-hand-dominant female diagnosed with Hajdu–Cheney Syndrome presented with the chief complaint of chronic right wrist pain that worsened with grip and loading activity. The patient had no history of trauma and had utilized a short-arm cock-up wrist brace without relief of her symptoms. The patient's medical history was consistent with polycystic kidney disease and previous surgical history included a posterior cervical fusion, skull reconstruction, and eyelid reconstruction.

The patient was 149 cm in height (4 ft 11 in.), of normal intelligence and was attending college at the time of our initial encounter. Range of motion of her right wrist was as follows: extension 70°, flexion 60°, radial deviation 45°, and ulnar deviation 0°. The flexion–extension arc of the elbow ranged from 20 to 120°, while pronation was 80° and supination was limited to 10°. Level II grip strength using a Jamar hand dynamometer (Sammons-Preston, Bolingbrook, IL) was 9 kg on the right side and 14 kg on the left side (64 %). The patient had a negative triangular fibrocartilage complex (TFCC) stress test, but was tender over the scapholunate ligament and exhibited significant carpal laxity bilaterally, in particular when performing Watson's shift test.

Radiographs of the elbow, forearm, and wrist demonstrated generalized bone dysplasia, bilateral radial head dislocation, 1-cm positive ulnar variance, and incongruity of the DRUJ bilaterally (Fig. 1).

The patient was diagnosed with a combined pathology including: ulnar impaction syndrome, longitudinal forearm

A. Bachoura · S. M. Jacoby (✉) · A. L. Osterman
The Philadelphia Hand Center, Thomas Jefferson University,
700 South Henderson Road, Suite 200, King of Prussia,
Philadelphia, PA 19406, USA
e-mail: sidneyjacoby@gmail.com

Fig. 1 **a** An AP view of the right wrist demonstrating positive ulnar variance, DRUJ dissociation, and carpal dysplasia. **b** An AP view of the right forearm demonstrating generalized skeletal dysplasia, marked positive ulnar variance as well as incongruity and dislocation of the DRUJ. **c** Lateral radiograph of the forearm demonstrating a laterally dislocated radial head

instability, and radial head dislocation. After detailed conversation with the patient, it was decided that one-bone forearm (OBF) surgery would result in the most predictable outcome with regard to pain relief and forearm function, but in the process would sacrifice forearm rotation.

The patient consented to the procedure and under axillary block augmented with sedation as well as standard pre-operative antibiotics, underwent wrist arthroscopy. The ulna head was observed to protrude through a large rent in the

central TFCC and there was marked ulnar synovitis. Synovectomy and debridement of the TFCC was performed. The arm was then removed from the distraction tower and the tourniquet inflated. An oblique incision across the dorsal forearm was made, starting at the proximal ulna and extending to the radius. Both bones were exposed supra-periosteally over an interval of approximately 8 cm. The interosseous membrane was observed to be lax and deficient, particularly the central band. Oblique osteotomies were performed first in the radius and then in the ulna. The bones were then reduced in end-to-end fashion with the hand in slight pronation, and a five-hole locking plate (Synthes, Paoli, PA) was applied using a combination of compression and locking screws to achieve stable fixation. The midshaft of the radius was also fixed to the newly constructed one-bone forearm using one lag screw. Concerns about injury to the interosseous nerves limited our options to dissect proximal to the proximal edge of the plate as shown in Fig. 2. Although AO principles of 6 cortices on either side of the osteotomy were not observed, it was possible to achieve stable arthrodesis with the construct utilized, which had four cortices, and a lag screw proximal to the osteotomy site. Intraoperative fluoroscopy confirmed reduction of the osteotomy and appropriate screw length. Ulnar variance was noted to be 2 mm negative at the wrist, and the wrist did not translocate ulnarly with stress testing. A separate lateral incision was then made proximally over the radial head, which was found to be dislocated posteriorly and subsequently excised with the use of an oscillating saw. The excised radial head was then morcellized and used as bone graft at the osteotomy site. The patient was provided with a protective wrist splint and underwent formal therapy that consisted of light active range of motion exercises of the fingers, wrist, and elbow for 2 months, followed by strengthening exercises for 1 month. Follow-up radiographs demonstrated bony union, and the patient experienced an uneventful postoperative course (Fig. 2).

The patient returned to clinic 28 months later complaining of right shoulder pain. She had full range of motion at the shoulder and elbow, wrist extension was 60°, wrist flexion was 55°, level II grip strength was 16 kg on the right and 20 kg on the left (80 %). A shoulder MRI was performed and found to be normal. The patient was advised that her pain was secondary to overuse following her OBF procedure and she was advised to modify her shoulder mechanics as instructed by our therapy team. Eight years following her surgery, the patient was contacted by telephone. Her Quick Disabilities of the Arm, Shoulder and Hand score was 21, she reported no upper extremity pain (0/10), she was completely satisfied with treatment (10/10) and had not undergone any additional procedures.

Fig. 2 **a** An AP radiograph of the forearm 6 months following OBF construction demonstrating bone union at the osteotomy site and negative ulnar variance. **b** Lateral radiograph of the forearm 6 months postoperatively. The radial head has been excised

Discussion

One-bone forearm surgery is generally regarded as one of the last available salvage procedures that could be used to treat patients with distal radio-ulnar joint (DRUJ), distal ulna stump, or longitudinal forearm instability secondary to a congenital, oncologic, or a post-traumatic primary etiology. The procedure is performed by arthrodesis of the proximal ulna with the distal shaft of the radius. In the process, forearm pronation and supination are sacrificed, but ulnohumeral and radiocarpal motions are preserved.

In general, superior functional and patient rated outcomes have been observed when OBF surgery has been used to treat forearm instability secondary to congenital or oncological disorders as compared to post-traumatic causes [5, 11]. Peterson CA et al. categorized OBF patients into two groups based on the etiology of injury: (1) post-traumatic; or (2) an oncologic or congenital etiology [11]. In their series, post-traumatic patients had undergone more previous surgeries, had a higher incidence of prior infection, presented with poorer soft tissue and bone stock, and had overall poorer results with higher complication rates than the non-trauma group. The authors deduced that the greater degree of initial insult to the forearm in post-traumatic cases predicated worse results regardless of reconstruction technique.

In specific cases of forearm pathology that involve an incompetent DRUJ and a dislocated radial head, Peterson HA suggested that immediate OBF construction would avoid further progressive radial head dislocation and futile surgery to reduce it [12]. Furthermore, he noted that the one-bone forearm is most optimally constructed and functions best when the proximal ulna and distal radius are initially present. Our results corroborate both of these findings as our

patient had only one definitive surgery and reported very good function and no pain many years after surgery.

It is important to note that Fujioka et al. reported a remarkably similar case of a patient with Hajdu–Cheney syndrome. The patient was a 46 year old female who presented with wrist pain and was found to have dorsal displacement of the distal ulna, positive ulnar variance, limited supination and extensor tendon rupture, generalized ligamentous laxity and incongruity of the radio-humeral joint [1]. The authors reconstructed the ruptured tendons, and managed the distal ulna dislocation with a Sauve–Kapandji procedure. The patient reported an improvement of her symptoms, and by 6 months, had no pain or limitations in the range of motion of her wrist or digits. Two years later however, the patient developed marked, but asymptomatic longitudinal forearm instability that resulted in anterior dislocation of the radial head with proximal migration of the radius. It is unclear how the patient's symptoms evolved thereafter. The advantage of OBF surgery is its potential to provide a more predictable postoperative course with respect to pain and stability, especially in younger and more active patients. In our case, the application of a seemingly radical surgical intervention as the first line and definitive form of treatment proved to be a viable option that provided the patient with acceptable upper extremity function, no pain, and complete satisfaction 8-years after surgery.

Patients that undergo OBF surgery usually compensate lost forearm rotation by a combination of shoulder and wrist hypermobility, which may lead to symptoms of overuse [12]. Therefore, patient education aimed at avoiding these sequelae should be elucidated preoperatively and during the early postoperative time period. Surprisingly, the patient described in this case report had a 10° loss of wrist extension and a 5°

loss of wrist flexion, despite the expected improvements that typically accompany compensatory adaptation, as well as ulna shortening [9]. One study however, found a decrease in wrist motion in the flexion–extension plane following ulna shortening osteotomy and the fact that our patient underwent ulnar shortening could explain in part, the slight decrease in motion noted [8]. Alternatively, the limitations of clinical measurements, which depend on the rigor and the methodology of the examiner, may explain the counter-intuitive results. Furthermore, passive versus active measurements, which were not specified in the medical record may have contributed to inconsistent measuring methodology and subsequent difficulties in interpretation, as patients with generalized ligamentous laxity would be expected to have considerably different active and passive joint motion.

In certain scenarios of DRUJ and distal ulna stump instability, the introduction of constrained and non-constrained DRUJ prostheses has provided a viable alternative to OBF surgery. These prostheses have been effective in preserving forearm rotation while simultaneously addressing painful DRUJ instability [7, 15]. The implants, however, cannot treat longitudinal forearm instability and are generally not recommended for patients with poor bone stock or osteoporosis. Furthermore, prosthetic replacement in young and active patients generally raises concerns about early implant failure and loosening. Therefore, in unique circumstances, once conservative measures have failed, and when less radical reconstructive procedures are not appropriate, OBF surgery remains a valuable salvage procedure, and is capable of providing a good postoperative course. Similar to Peterson CA et al. [11], in our experience, the OBF procedure is most effective in young patients with congenital or genetic defects.

Statement of human and animal rights The Thomas Jefferson University Institutional Review Board approved the study protocol, which was conducted accordingly under its guidelines.

Conflict of interest The authors declare no conflicts of interest related to the material presented in this report.

Statement of informed consent Informed consent was obtained from the participant described in this report.

References

1. Fujioka H, Tanaka J, Yoshiya S, et al. Proximal translation of the radius following arthroplasty of the distal radioulnar joint in Hajdu–Cheney syndrome. *J Shoulder Elbow Surg.* 2003;12:97–100.
2. Herscovici Jr D, Bowen JR, Scott Jr CI. Cervical instability as an unusual manifestation of Hajdu–Cheney syndrome of acroosteolysis. *Clin Orthop Relat Res.* 1990;255:111–6.
3. Hilton MJ, Tu X, Wu X, et al. Notch signaling maintains bone marrow mesenchymal progenitors by suppressing osteoblast differentiation. *Nat Med.* 2008;14:306–14.
4. Iwaya T, Taniguchi K, Watanabe J, et al. Hajdu–Cheney syndrome. *Arch Orthop Traum Surg.* 1979;95:293–302.
5. Jacoby SM, Bachoura A, DiPrinzio EV, et al. Complications following one-bone forearm surgery for posttraumatic forearm and distal radio-ulnar joint instability. *J Hand Surg Am.* 2013;38:976–982.e1.
6. Kaplan P, Ramos F, Zackai EH, et al. Cystic kidney disease in Hajdu–Cheney syndrome. *Am J Med Gen.* 1995;56:25–30.
7. Laurentin-Pérez LA, Goodwin AN, Babb BA, et al. A study of functional outcomes following implantation of a total distal radioulnar joint prosthesis. *J Hand Surg Eur Vol.* 2008;33:18–28.
8. Loh YC, Van Den Abbeele K, Stanley JK, et al. The results of ulnar shortening for ulnar impaction syndrome. *J Hand Surg Br.* 1999;24:316–20.
9. Minami A, Kato H. Ulnar shortening for triangular fibrocartilage complex tears associated with ulnar positive variance. *J Hand Surg Am.* 1998;23:904–8.
10. O'Reilly MA, Shaw DG. Hajdu–Cheney syndrome. *Ann Rheum Dis.* 1994;53:276–9.
11. Peterson CA 2nd, Maki S, Wood MB. Clinical results of the one-bone forearm. *J Hand Surg Am.* 1995;20:609–18.
12. Peterson HA. The ulnius: a one-bone forearm in children. *J Pediatr Orthop B.* 2008;17:95–101.
13. Simpson MA, Irving MD, Asilmaz E, et al. Mutations in NOTCH2 cause Hajdu–Cheney syndrome, a disorder of severe and progressive bone loss. *Nat Genet.* 2011;43:303–5.
14. Stathopoulos IP, Trovas G, Lampropoulou-Adamidou K, et al. Severe osteoporosis and mutation in NOTCH2 gene in a woman with Hajdu–Cheney syndrome. *Bone.* 2013;52:366–71.
15. van Schoonhoven J, Mühldorfer-Fodor M, Fernandez DL, et al. Salvage of failed resection arthroplasties of the distal radioulnar joint using an ulnar head prosthesis: long-term results. *J Hand Surg Am.* 2012;37:1372–80.