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Total shoulder arthroplasty in a patient with microcephalic osteodysplastic dysplasia: a case report with 4-year outcomes

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Microcephalic osteodysplastic dysplasia is an extremely rare genetic condition that is similar to osteodysplastic primordial dwarfism types I to III and Seckel syndrome, but with few distinguishing features.[4,7] Primary distinguishing characteristics include vertebral body irregularity, odontoid process and first lumbar vertebra hypoplasia, syrinx of the spinal cord, short metacarpals and metatarsals, coned epiphyses, and coxa valga. This condition is associated with normal cognitive development, allowing affected patients to reach adulthood.[4] We present a case report of a 21-year-old woman with microcephalic osteodysplastic dysplasia who was treated with right total shoulder arthroplasty. To our knowledge, total shoulder arthroplasty associated with this rare genetic condition has not been previously described in the literature.

Case report

A 21-year-old left hand–dominant woman with microcephalic osteodysplastic dysplasia presented with progressively intensifying right shoulder pain that had become highly debilitating, limiting daily activity, shoulder range of motion, and contributing to considerable night pain. The patient was a college communications major and worked part-time at a department store as a cashier at the time of surgery.

Associated with the genetic condition, the patient presented with short stature, a prominent forehead and chin, bilateral clubfeet, scoliosis, brachydactyly, and a history of bilateral radial head subluxations. Her surgical history included cervical vertebrae 1-2 fusion for os odontoideum in 2002, tethered spinal cord release in 1990, cataract surgery, and bilateral clubfoot correction. At the time of surgery, she was taking Fosamax (Merck & Co, Inc, Whitehouse Station, NJ, USA) and 1000 units of vitamin D_3 daily.

The initial orthopedic evaluation found severe right glenohumeral joint degenerative changes with a chief complaint of night pain and mechanical locking. On the basis of the severe pain, intermittent glenohumeral joint locking, and inability to effectively perform essential activities of daily living, the decision was made to perform right total shoulder

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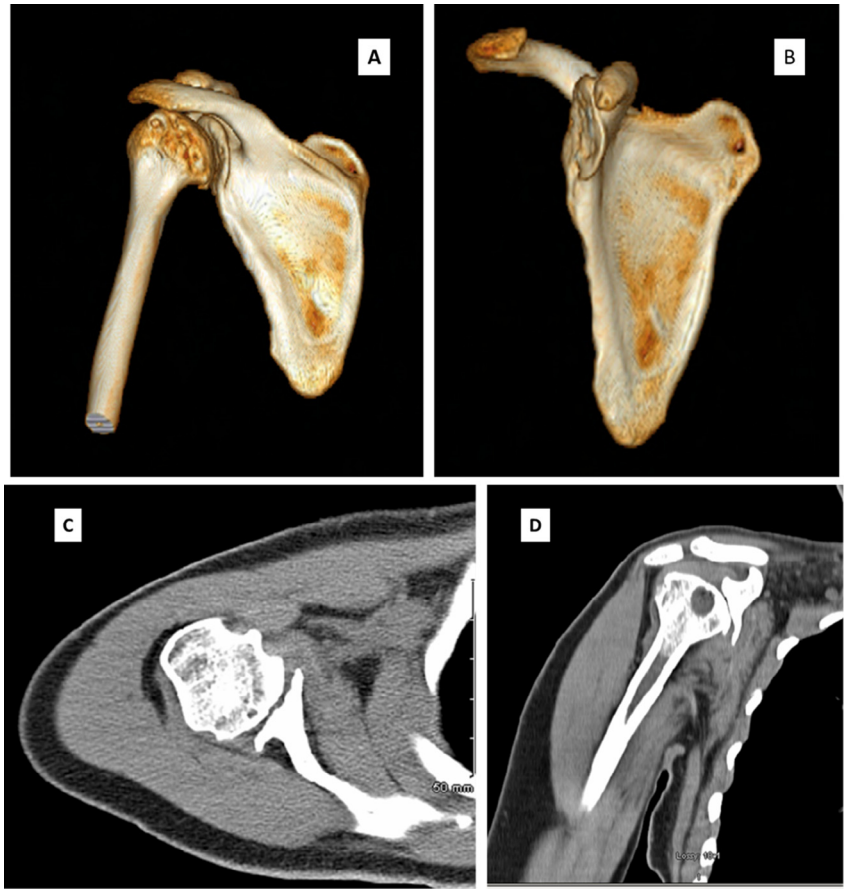

Figure 1 Presurgery 3-dimensional (3D) reconstructions demonstrate (**A**) significant degenerative changes at the proximal humerus and glenoid process and (**B**) show the glenoid process with severe glenoid wear. A presurgery computed tomography scan (**C**) axillary view demonstrates bony erosion, increased retroversion, and small glenoid process size, and a (**D**) coronal view demonstrates bony erosion, increased retroversion, and small glenoid process size.

arthroplasty. In addition to follow-up orthopedic examinations, the SF-12, the American Shoulder and Elbow Surgeon (ASES) Surgeon Score, and ASES Patient Self-Evaluation Scores were completed before surgery and at more than 4 years after surgery.[6,9] The patient also completed the 11-item version of the Disabilities of Arm, Shoulder and Hand Outcome Measure before surgery, 1 week after surgery during her initial physical therapy evaluation, at discharge from physical therapy, and at 4 years.[1,5]

The preoperative evaluation revealed she was 109.2 cm (43 inches) tall and weighed 24.9 kg (55 pounds), and her orthopedic examination findings were consistent with her history. Focused upper extremity examination revealed tenderness to palpation along the anterior and posterior joint line of the right shoulder. There was crepitus, decreased strength, and limited active and passive range of motion on the right compared with the left shoulder. Right shoulder range of motion and strength deficits were associated with pain. Neurovascular examination of the right upper extremity was grossly intact, and reflexes were normal. There were no findings to suggest rotator cuff tear or impingement.

Right shoulder computed tomography (CT) scans taken during the initial office visit demonstrated significant degenerative changes in the proximal humerus and glenoid process, increased retroversion, small glenoid and humeral head size, severe glenoid wear, severe glenohumeral joint arthropathy, and a large proximal humeral cyst (Fig. 1). There was no evidence of a rotator cuff tear; however, results of clinical impingement tests suggested tendinopathy.

After thorough discussion of operative and nonoperative treatment options with the patient and her family, she made the decision to undergo right total shoulder arthroplasty. Because of this patient's unique anthropometric characteristics, custom fabrication of a total shoulder arthroplasty implant was a necessity.

A custom prosthesis with corresponding instruments were designed in conjunction with engineers based on preoperative imaging, which included standard 3-view radiographs (anteroposterior, scapula Y, and axillary views) and CT scans. These CT scan images enabled the creation of 3-dimensional (3D) models to effectively match implant design with the patient's unique bony architecture. These models helped