Spinal reconstruction in Hajdu-Cheney syndrome

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Hajdu-Cheney syndrome (HCS), or acro-osteolysis, is described by the National Organization for Rare Disorders (NORD) as a rare, heritable disorder of connective tissue. A major characteristic of HCS is breakdown of the bone (osteolysis), especially the outermost bones of the fingers and toes (acro-osteolysis). Patients with this syndrome also frequently have skull deformities, short stature, joint laxity, and osteoporosis. A small proportion show mild mental retardation, but most affected persons have normal mental development.1

According to NORD, only about 50 cases of HCS have been reported in the medical literature. This article reports follow-up to a case that was reported by Elias and colleagues in 1978.2 They described a mother and son with generalized osseous dysplasia. Both the mother and son were described as being of short stature with low-set ears, bushy eyebrows, a broad nose without hypertelorism, and a receding chin. Pseudoclubbing of the nails was present along with shortening of the distal phalanges. The fourth finger of the left hand of the son had been spared at the time of the Elias article. At that time, the boy was 12 years old.2 In this report, we describe our recent experience with reconstruction of the cervical spine on the son, who is now 42 years old.

CASE

This 42-year-old male presented to our institution’s emergency department after sustaining injuries as the driver of a truck that had collided with a tree. He was not wearing a

**FIGURE 1.** A reconstructed midline sagittal CT image taken before surgery shows kyphotic deformity and multiple wedge compression fractures of C5, C6, and C7.

**FIGURE 2.** CT taken after surgery demonstrates the C5-T1 anterior fusion with dynamic anterior locking plate and contoured patella allograft. Sagittal alignment has improved.
The Surgical Patient

seat belt. He had experienced loss of consciousness and complained of upper extremity paresthesia.

On physical examination, he was found to have a forearm laceration, which was repaired, and no neurologic deficit. Of note were the pseudoclubbing of his nails and the shortening of the distal phalanges, including the left fourth finger that had been previously spared. Imaging studies showed fractures at C5-6 and C7-T1 with a kyphotic deformity of the cervical spine (see Figure 1, page 29). The patient also had what appeared to be perched facets at C6-7.

He was taken to the operating room for a deformity correction and stabilization procedure. Because of his significant osteoporosis, a more aggressive reconstructive approach was needed to spread the forces over many levels. For this reason, a staged procedure was planned with a back/front/back approach to the correction.

The first stage of the procedure was a C5-T4 posterior release with osteotomies at C6 and C7. Attempts were made at reduction but were not successful because of the anterior wedge fractures. Lateral mass screws were placed in preparation for later placement of the posterior rod that would augment the anterior portion of the fusion construct. At this stage, the rod was not attached because further deformity correction was required anteriorly. Because the patient had tolerated the procedure well thus far and it had moved along quickly, the next stage was immediately initiated.

At the second stage of the surgery, anterior arthrodesis of C5-T1 was performed, but with some difficulty because of the patient’s obesity. Deformity correction and stabilization was achieved by complete diskectomies at C5-6, C6-7, and C7-T1 and insertion of contoured patella allograft and anterior locking plate. Throughout the anterior portion of the procedure, the softness of the patient’s bone required careful selection of the implants to maximize bone implant interface.

The final stage of the procedure was then completed because the patient had continued to do well during the first two stages. The posterior approach was reopened, and the posterior rod was attached to the screws that were placed in the first stage. Crushed cancellous allograft with a small amount of local autograft was mixed with demineralized bone products. No bone morphogenic protein (BMP) was utilized for fusion augmentation. The final deformity correction was acceptable (see Figure 2, page 29). The patient had tolerated the entire procedure well and was taken to the recovery room in good condition.

Complications occurred during the postoperative period, starting with extubation difficulty. Within the first week, an anterior incision wound infection developed. The patient also sustained a non-ST elevation MI that was complicated by a cerebrovascular embolic event. He subsequently had an excellent neurologic recovery and, after a stay in a rehabilitation unit, was discharged home. At discharge, radiographs showed that he maintained the deformity correction with his hardware in good position. While at home,
he was noncompliant with rehabilitation instructions by not wearing his cervical orthosis at all times.

The neurosurgery service was consulted again when the patient was readmitted to the hospital because of respiratory stridor. Though he had no neck pain, routine follow-up imaging of the cervical spine revealed fusion failure and loss of correction (see Figure 3, page 31). The patient was returned to the operating room for a posterior-only revision with positional deformity correction and instrumented fusion from C2 through T4 (multi-axial lateral mass/pedicle screw rod construct) (see Figure 4, page 31). BMP augmented the autograft and allograft bone. After more vigorous compliance with the wearing of the cervical orthosis, the patient went on to full fusion with maintenance of good sagittal balance both locally and globally. He remained without neck pain and neurologically intact, though he required long-term tracheotomy because of pre-existing tracheal stenosis.

**DISCUSSION**

In 1948, Hajdu and Kauntze described a patient with “basil impression, cranio-facial and peripheral dysostosis along with spinal osteoporosis and chondrodystrophy.”3 In 1965, Cheney reported on a family with similar features, establishing the dominant genetic component of this syndrome.4

Greek word *acros* in this syndrome refers to the tip of the extremities. The word *osteolysis* refers to bone disintegration. *Acro-osteolysis* thus refers to the disintegration of bone specifically at the tips of the phalanges.4

Although HCS is well described in the literature, there are few reports of spinal surgery in these patients. In 1984, Chodoroff and colleagues reported their rehabilitation case experience with a 25-year-old female who had undergone decompression of the cervical spinal cord.5 This patient underwent a posterior fossa decompression with a C1 arch laminectomy with posterior cervical fusion and was subsequently placed in a halo traction vest postoperatively. We know nothing about the patient’s subsequent outcome.

Herscovici and colleagues describe a case of HCS in a 7-year-old female with progressive instability of the cervical spine, which required a spinal fusion with bone graft from C1-C3.6 Because of resorption of the bone graft, the patient was readmitted 1 year later for revision of the fusion mass. She required another hospitalization at the age of 18 years when she was found to have cervical instability of the C3-C4 level. At that time, she underwent an occiput to C6 posterior fusion using iliac crest supported by crushed donor graft. Follow-up radiographs revealed osteolysis of the fusion mass.6 Apart from these, there are no reports of surgical intervention of the spine in patients with HCS.

In 2001, Brennan and Pauli summarized the changing physical findings and medical sequelae over time in patients with HCS.7 In their analysis, 60% of young adults with this syndrome have spinal abnormalities secondary to bone absorption. “Vertebral compression, compression fractures and biconcave ‘fishbone’ deformities” were present in 28 of the 57 patients they studied. Kyphosis was demonstrated in 23% of the patients, scoliosis was found in 37%, and both kyphosis and scoliosis were found in 14%. Fifty-six percent of those with decreased bone density were found to have vertebral compression fractures.7

Our case raises a question about the ability of a patient with HCS to assimilate allograft/autograft bone and fuse. Bone grafting is used to stimulate and promote bone growth in order to develop a fusion mass, and BMP technology may be able to stimulate a better fusion process. Considering BMP’s ability to biologically stimulate capillary, perivascular tissue, and osteoprogenitor cell ingrowth to affect desired bone formation, persons with HCS may be a subcategory of patients who would benefit from its use.8,9

In our patient, we used an allograft of patella and demineralized bone matrix in the first operation, with subsequent fusion failure. In the revision surgery, we used BMP with autograft/allograft bone, crushed cancellous bone mixed with demineralized bone matrix. In animal fusion models, direct current electrical stimulation showed an increase in osteogenic genes and an increase in the healing rate and strength of bone,10,11 so we also placed a bone stimulator on the patient after his revision. This approach was effective.
and 6 months after the revision surgery, the fusion was stable (see Figure 5).

The patient and his wife were given significant education about the appropriate use of a cervical collar. Effective immobilization, positioning, and stability are necessary for effective healing and fusion. A myriad of cervical orthotic devices are available; in our practice, we generally use the Miami J or Aspen collar. Askins and Eismont have shown the Miami J to be a superior orthotic for restricting motion of the cervical spine.12

Final comment Although HCS is a rare condition, patients who have it are likely to find themselves in need of corrective spinal surgery. The ongoing osteolysis in these patients means that understanding the disease process will be an important consideration for the spine surgeon. The added expense of additional implants, BMP, bone stimulators, and aggressive orthosis is more than offset by the decreased need for revision surgery. Our case represents the intense correction that may be needed for a patient with this syndrome. JAAPA

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REFERENCES