Surgical management of Hirayama disease in a pediatric patient presenting with severe cervical kyphosis and focal myelopathy: illustrative case

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BACKGROUND Hirayama disease (HD) is a rare, nonfamilial neuromuscular disease causing cervical myelopathy and deformity, most commonly affecting pubertal Asian males. Patients whose nonoperative treatment fails and who cannot tolerate long-term cervical immobilization, experience relapse after arrest of symptoms, or present with severe features warrant surgical treatment. Here, the authors present an unusual case of HD that resulted in rapid progression of severe cervical kyphosis and discuss surgical management strategies.

OBSERVATIONS A 15-year-old male presented with unprovoked neck pain, progressive chin-on-chest phenomenon, and cervical myelopathy. Imaging revealed a severe subaxial cervical kyphosis of 88° and severe spinal cord compression secondary to changes within the thecal sac, ligaments, and bony elements. He underwent a multistage surgery involving halo gravity traction, C3–6 anterior cervical discectomy and fusion, and C2 to T2 posterior instrumented fusion with C3–5 Smith-Petersen osteotomies. Cervical subaxial pedicle screws facilitated deformity correction through a cantilever technique.

LESSONS HD is rare and often self-limited. For severe or refractory cases of HD, guidelines for surgical management have been suggested, with a variety of approaches deemed efficacious. This is the first case of a patient presenting with such severe cervical deformity; early diagnosis and recognition is the first step toward prompt, adequate management.

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KEYWORDS case report; cervical deformity; cervical myelopathy; deformity; Hirayama disease; kyphosis; monomelic amyotrophy

Hirayama disease (HD) also, known as monomelic amyotrophy, is a rare, nonfamilial cervical myelopathy characterized by asymmetrical distal upper extremity weakness.1 Most commonly affecting males of Asian descent, it presents with insidious onset during puberty.2 HD is hypothesized to manifest from ischemic injury to cervical anterior horn cells as a result of abnormalities in dorsal dural attachments and thecal sac hypermobility.3–5 Although the mechanism underlying this process is unclear, it may stem from a mismatch between the spinal cord and canal lengths during growth spurts in combination with weak neck extensors.6–8 Magnetic resonance imaging (MRI) reveals cervical straightening or mild kyphosis, flattening of the midlower cervical cord, and a crescent-shaped posterior epidural space on T2-weighted imaging.5,9 Electrodagnostic studies reveal normal or mildly impaired conduction velocities and classical sparing of the cranial, thoracic, lumbar, and sacral nerves.1,2,10

Although often self-limited, HD has the potential to lead to progressive cervical myelopathy and concomitant disability.1 Many advocate for nonoperative management of HD,11 but surgical intervention may be unavoidable in a subset of patients.12 Here, we present an unusual case of HD that resulted in rapid progression of severe cervical kyphosis, and we discuss surgical management strategies. This case report has been reported in line with the SCARE (Surgical Case Report) criteria.13

Illustrative Case

History and Examination

A 15-year-old male presented with 1 year of unprovoked abnormal head position and mechanical neck pain that improved with recumbency and cervical immobilization. On examination, he had a
chin-on-chest phenomenon and asymmetrical hand grip and interossei weakness, left greater than right, along with spasticity in his extremities and atrophy of intrinsic muscles. Imaging revealed an 88° kyphotic deformity centered at C4–5, with a cervical sagittal vertical axis (cSVA) of 3.4 cm and splayed facets from C3 to C6 (Fig. 1). Draping of the ventral cord over the deformity apex; cord compression with T2 signal abnormality; and an expanded, crescent-shaped posterior epidural space were evident on MRI (Fig. 1E). Flexion-extension MRI was avoided, given the severity of his deformity and the presence of neurological deficits. Electrodiagnostic testing was deferred because the results would not have influenced surgical management. The diagnosis of HD was therefore made on the basis of the occult onset of his disease during growth, asymmetrical upper extremity weakness, and MRI findings.

Interventions and Outcome

The patient underwent staged treatment, beginning with halo gravity traction (HGT) incremented up to 25 pounds over 5 days (Fig. 2). Subsequently, he underwent anterior release via C3–6 anterior cervical discectomies and fusions (ACDFs), as well as uncovertebral joint resections, with standalone interbody cages affixed only to the superior endplate (Fig. 2B). This was followed by HGT for 5 days. Last, he underwent posterior instrumented fusion from C2 to T2, with C3–6 laminectomies and Smith-Petersen osteotomies (Fig. 3). Pedicle screw fixation was employed on the left side, facilitating deformity correction through a cantilever technique with a 4.0-mm cobalt chrome rod.

Motor evoked potentials were briefly lost during decompression and returned with the elevation of mean arterial pressure (MAP) to 90 mm Hg. Upon emergence from surgery, his neurological examination was at baseline, but he developed left hemiparesis 4 hours postoperatively. Steroids were initiated, and his MAP increased to greater than 100 mm Hg. Emergent imaging ruled out hematoma, infarct, or hardware complications. His examination recovered to baseline by the following morning, and MAPs were weaned over 4 days without complication. He was ultimately discharged on postoperative day 5. Scoliosis radiographs demonstrated an cSVA of 1.7 cm and cervical kyphosis of 2° (86° of correction) at the 3-month follow-up.

Discussion

Observations

HD is a rare disease with a low coincidence of deformity. To our knowledge, this is the first case of a patient presenting with chin-on-chest phenomenon. Presenting symptoms of HD typically arrest between 2 and 5 years after onset.1,12 Nonetheless, approximately 70% of patients have persistent disabilities,14 and early intervention can limit disease progression and serious disability.12,15–17

Cervical immobilization to limit flexion until growth spurts are completed or until arrest of symptoms is observed is the mainstay of early treatment.1,10,11,18,19 Physiotherapy to strengthen cervical extensors is also recommended.1,6,12 Consensus guidelines also support trialing neurotrophic pharmacological treatment in patients with HD.12,20

Patients whose nonoperative treatment fails and who cannot tolerate long-term cervical immobilization, experience relapse after arrest of symptoms, or present with severe features warrant consideration for operative management.1,12 The aims of surgery are to decompress the spinal cord and eliminate venous engorgement of the posterior epidural venous plexus.12,21 Various surgical approaches to treat HD have been described: laminectomy, laminoplasty, laminectomy with instrumented

FIG. 1. Preoperative anteroposterior (A) and lateral (B) standing scoliosis radiographs showing cervical kyphosis of 88° and cSVA of 3.4 cm. Flexion (C) and extension (D) radiographs showing rigid deformity. Midsagittal MRI showing T2 cord signal change at the apex of the kyphotic deformity (E).

FIG. 2. Lateral radiographs showing reduction of the kyphotic deformity to 21° following HGT (A) and to 13° following C3–6 ACDF, with the patient maintained in traction (B).
fusion, and ACDF. A recent meta-analysis found no significant difference in the pooled outcome of different surgical approaches. As such, the surgical approach should be tailored to the specific needs of the patient.

**Lessons**

The case presented is unique due to the severity of deformity. HGT is a powerful approach allowing maximal deformity correction and acclimatization of the spinal cord to potential stretch or manipulation. Anterior releases alone cannot adequately address severe subaxial kyphoses and were thus employed as a preparatory supplement to the posterior correction in this case. As shown, even after anterior release, 13° kyphosis remained.

Subaxial pedicle screw fixation provides four times the pullout strength of lateral mass fixation, offering advantages in cervical deformity surgery. Enabling technologies, such as navigation, have made placement much more accurate and safer. Here, pedicle screw fixation allowed aggressive rostral to caudal cantilever correction using a patient-specific cobalt chrome rod that was designed preoperatively.

Achieving adequate deformity correction and decompressing the spinal cord are equally important in HD. In the first studies evaluating cervical sagittal alignment in patients with HD, authors have reported correction of modest abnormalities in cervical lordosis and sagittal alignment following surgery. Other authors have correlated cervical sagittal balance indices with clinical outcomes following surgery for HD. Finite element modeling predicts that factors predisposing to adjacent segment disease are minimal if cervical lordosis is maintained in patients with HD, underscoring the importance of considering correction of spinal deformity when surgery is indicated.

In summary, HD is uncommon and often self-limited. For severe or refractory cases of HD, guidelines for surgical management have been developed, with a variety of approaches deemed efficacious. It is unusual for a patient with HD to present with severe cervical deformity, and this is the first report detailing operative management to address such severe pathology.

**References**


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