Intraosseous CSF Fistula in a Patient with Gorham Disease Resulting in Intracranial Hypotension

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SUMMARY: A 7-year-old girl with a history of headaches and Gorham disease was surgically treated in infancy for Chiari I malformation. Subsequent investigation revealed that her cerebellar tonsillar ectopia was due to a long-standing spinal CSF-lymphatic fistula causing intracranial hypotension. Percutaneous fistula closure was performed several times, resulting in transient symptomatic improvement.

ABBREVIATION: n-BCA = n-butyl cyanoacrylate

Gorham disease, or massive osteolysis, is a rare disorder characterized by proliferation of lymphatic channels with resulting bone destruction. Chylous pleural and pericardial effusions from extension of osseous disease into the pleural space and mediastinum and 1 case of a CSF-pleural fistula have been reported.1-4 Chiari I malformation has also been reported in 7 patients with Gorham or related diseases, though the relationship between the 2 conditions is not clear.5-8 We describe a child with Gorham disease who was initially treated for Chiari I malformation, which was, in retrospect, secondary to intracranial hypotension from a CSF fistula between a lumbar nerve root sleeve and a lymphatic malformation.

Case Report
A 1-year-old girl with no other medical history presented with crying and head holding. MR imaging demonstrated low cerebellar tonsils consistent with a Chiari I malformation, along with cervical cord syrinx. The patient underwent posterior fossa decompression and duraplasty, with improved but persistent symptoms (Fig 1A).

Spine MR imaging performed to evaluate the syrinx showed a permissive osteolytic process involving the entire lumbosacral spine and pelvis, characterized by fatty marrow replacement. For several years, serial MR imaging revealed progressive replacement of bone with fluid, consistent with Gorham disease (Fig 1B, -C). The patient was treated with interferon α 2b and zoledronate (Zometa) for 1 year, with stabilization of her imaging findings. She continues to take oral vitamin D, calcium supplementation, and oral alendronate sodium (Fosamax).

Worsening postural headaches prompted a repeat MR imaging at age 7, which showed features of intracranial hypotension, including tonsillar descent, brain stem sag, draping of the floor of the third ventricle over the dorsum sellae, and diffusely enlarged dural venous sinuses. In retrospect, several of these features were also present at age 1.

A CT myelogram demonstrated rapid egress of contrast from the right L4 nerve root sleeve into the expanded L4 and L5 vertebral bodies, ultimately spreading into the paraspinal soft tissues and retroperitoneum (Fig 2A, -B). The patient underwent 3 CT-guided transforaminal blood patches (10–20 mL) directed at the fistula, each with transient improvement of headaches. For each procedure, a 25-ga spinal needle was positioned within the thecal sac at L3, with the patient in a prone position, and rapid CT was performed during infusion of 5 mL of iohexol contrast (Omnipaque-240; GE Healthcare, Princeton, New Jersey). The needle was left in the thecal sac to monitor treatment progress.

Follow-up MR imaging showed improvement in findings of CSF hypotension. Because of recrudescence headaches and persistence of the fistula, CT-guided placement of 0.8 mL of n-BCA liquid embolic into the receiving cavity of the fistula was performed, with documentation of reduction of flow through the fistula (Fig 2C). Several days following treatment, the patient’s headaches significantly improved. This improvement lasted 6 months, but she continues to have mild positional headaches and evidence of intracranial hypotension on MR imaging.

Discussion
The pathophysiology of Gorham disease, also known as “vanishing bone disease” or “massive osteolysis,” remains poorly understood. In the early stage, resorbed bone is replaced by non-neoplastic hypervascular angiomatous and fibrous connective tissue, with proliferative vessels showing either capillary, sinusoidal, or cavernous histology. In the later stages, progressive dissolution of bone occurs, with replacement by fibrous tissue.1-3,9 Although the exact nature of the disease remains unknown, the effects of hyperemia, changes in local pH-stimulating hydrolytic enzyme activity, and increased osteoclast activity have all been proposed as contributing factors.10

Gorham disease occurs in men or women of any age group but is usually discovered before 40 years of age. It may involve any bone within the axial or appendicular skeleton, though spine and visceral involvement have higher morbidity and mortality.1,3,4 Chylothorax from accumulation of lymph or thoracic duct disruption may also be associated with significant morbidity.1,3,4 Bone destruction tends to be regional, unimpeded by joint or disk spaces, and progressive. Medical treatment options include antosteoclastic agents (bisphosphonates), radiation therapy, and interferon α 2b.10,11 Although resection has been attempted in localized disease, no single therapy is effective. MR imaging typically shows re-
placement of normal bone marrow with high T1 signal intensity from fat. In our case, very high signal intensity within lumbar vertebral bodies was also visible on T2-weighted images due to CSF accumulation from the fistula.

Cerebellar tonsillar ectopia in our patient was a manifestation of intracranial hypotension caused by a high-volume lumbar CSF fistula, with syrinx formation secondary to foramen magnum obstruction. Chiari I malformation has been described in 5 other patients with Gorham disease and in 2 with similar conditions. Agrawal et al reported a duropleural fistula in a patient with Gorham disease with a Chiari I malformation, but a potential relationship was not discussed. The likely etiology of the CSF fistula in our patient is similar to that proposed in previously reported cases: dural erosion by the angio proliferative process in the adjacent bone.

In any patient with suspected Chiari I malformation, MR images should be examined before decompression for signs of intracranial hypotension, such as sagging of posterior fossa structures, inferior displacement of the third ventricle, dural venous sinus enlargement, and diffuse dural enhancement. These findings should prompt an investigation to identify the source of the CSF leak. Direct treatment of the leak is the appropriate step, rather than suboccipital decompression surgery. The cumulative treatment effects of repeat blood patching and embolization in this case were sufficient to improve imaging findings of CSF hypotension and symptoms. Percutaneous injection of autologous blood or liquid embolics such as n-BCA can be performed safely under low-radiation-dose CT guidance. Laminectomy or fusion was not attempted due to fear of spinous instability or vertebral collapse.
References