Letter: Vanishing Act: Gorham-Stout disease leading to cerebrospinal fluid dynamic abnormalities

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We read with great interest the article by Stroh et al (1) describing neuro-ophthalmic involvement in a patient with Gorham-Stout disease. We present a patient with Gorham-Stout disease highlighting the potential cerebrospinal fluid (CSF) dynamic abnormalities that may occur in such patients.

A 14-year-old girl with no significant past medical history was evaluated by an otolaryngologist for progressive right-sided hearing loss. A right supraclavicular mass was detected and aspirated, but the pathology was non-diagnostic. CT of the temporal bones and neck revealed a large trans-spatial multiloculated hypodense cystic lesion in the right side of the neck, an ill-defined hypodense soft tissue mass with cortical erosion involving the right petrous temporal bone, and multifocal lytic expansile lesions with cortical dehiscence and osteolysis of the right mandible. (Figs 1). These findings were suggestive of a lymphatic malformation within the neck with associated osseous lymphangiomatosis in the spectrum of Gorham-Stout disease.

At the age of 18 years, she developed bacterial meningitis, which was treated and subsequently recurred. A CT cisternogram confirmed a CSF leak through the right temporal bone. MRI of the skull base confirmed severe osteolysis of the right mandible. The CSF leak was not surgically repaired at that time and the patient was treated with zoledronic acid in an effort to prevent further bone resorption, and an attempt to restore bone loss (2, 3).

MRI of the brain performed at age 19 years showed brainstem sagging, effacement of the suprasellar cistern, and low lying cerebellar tonsils (Fig 2). This combination of findings was
consistent with intracranial hypotension. MRI of the brain performed at age 20 years showed partial remineralization of the right mandible.

At age 20 years, the patient developed right otitis media. Due to concern that this might lead to another recurrence of meningitis, she was treated with ceftriaxone, cefdinir, and azithromycin. She then developed acute onset of severe headaches, neck pain, intermittent blurry vision, and binocular diplopia. Neuro-ophthalmic examination revealed visual acuity by 20/20 bilaterally, normal color vision and pupillary reactions, full extraocular movements but a 6 prism-diopter esotropia in right gaze and a 2 prism-diopter esophoria in left gaze. Ophthalmoscopy revealed bilateral optic disc edema (Fig 3). MRI of the brain demonstrated decreased effacement of the preopontine cistern, decreased brainstem sagging and cerebellar tonsillar descent compared to her previous MRI (Fig 4). MRV showed narrowing of both transverse and sigmoid venous sinuses. On lumbar puncture opening pressure was >56cm H$_2$O with 1445 RBCs/ml, 1 WBC/ml and normal protein and glucose levels and negative cultures. Despite the use of acetazolamide (500 mg bid) the patients sixth nerve palsies and optic disc edema worsend.

A lumbar drain improved her signs and symptoms, but once the lumbar drain was removed, her clinical examination worsened. Bilateral optic nerve sheath fenestrations were performed followed by placement of a ventriculoperitoneal shunt. One month later, her visual acuity was 20/20 in each eye, there was a 0.6 log unit right relative afferent pupillary defect, with a 2 prism-diopter esotropia in primary position and resolving optic disc edema bilaterally.

Bony involvement of the skull base in Gorham-Stout disease has been reported to lead to CSF leak and resulting CSF hypotension. Nazarian et al (4) reported a child with cranial lyphangiomatosis who developed recurrent meningitis and CSF otorrhea. Cushing et al (5) published a case of CSF leak due to Gorham-Stout disease involving the petrous apex in a 12-year old boy. Hernández-Marqués et al. (6) reported involvement of the temporal bone in a 2-year old boy leading to CSF otorrhea and secondary meningitis. Morimoto et al. (7) described an 11-year old girl with Gorham-Stout disease involving the right petrous apex that presented with elevated intracranial pressure (60cm H$_2$O on lumbar puncture) and CSF leak. The authors proposed that increased lymph entering the CSF space led to elevated intracranial pressure. CSF leak and intracranial hypotension from Gorham-Stout disease also have been described with lesions in the thoracic spine (8).

We propose that treatment with zolendronic acid in our patient led to reversal of bone loss and closure of the CSF leak and rebound intracranial hypertension. Our case expands the possible causes of CSF dynamic abnormalities which can occur in patients with Gorham-Stout disease.

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Abbreviations

CT  Computed tomography
MRI  Magnetic resonance imaging
CSF  Cerebrospinal fluid

References

Figure 1.
A. Postcontrast axial CT of the neck demonstrates a multiloculated cystic lesion on the right (arrows) and an expansile lesion in the right mandible with cortical dehiscence (arrowhead)
B. Coronal CT (bone algorithm) shows cortical erosion (arrow) involving the right temporal bone.
Figure 2.
Sagittal T1 MRI reveals effacement of the suprasellar and prepontine cisterns and low-lying cerebellar tonsils (arrow).
Figure 3.
Bilateral papilledema is present.
Figure 4.
Sagittal T1 MRI shows improvement in the position of the brainstem with less effacement of the suprasellar and prepontine cisterns and near normal position of the cerebellar tonsils.