Gorham-Stout Disease of the Temporal Bone
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Abstract

Objectives: 1) Present data regarding Gorham-Stout disease as manifested in the temporal bone. 2) Review the literature regarding this rare bone disorder.

Methods: A case report from a tertiary university hospital is presented. A literature search using the terms “Gorham-Stout disease and head and neck,” “massive osteolysis,” and “temporal bone” was undertaken.

Results: A 29-year-old female with a diagnosis of Gorham-Stout disease was identified. She complained of aural fullness and tinnitus bilaterally. Demineralization and moth-eaten changes of the osseous structures of the skull base and posterior fossa were prominent. The left mastoid air cells were opacified and erosion extended to the left jugular foramen, left hypoglossal canal, left stylo mastoid process, and left Eustachian tube. The histopathology and radiographic findings are presented.

Conclusion: Gorham-Stout disease is a rare disease of the bone and is also known as massive osteolysis. Less than 200 cases have been reported in the worldwide literature. This disease follows a rapidly progressive course resulting in resorption and replacement of bone with dense fibrous tissue. It typically presents in childhood or adolescence. Involvement within the head and neck is frequent but typically involves the calvarial bones. Involvement of the temporal bone is very rare. A review of the literature with a focus on head and neck manifestations of the disease and their management is presented.

Introduction

• Gorham-Stout Disease (GSD) is a rare bone disorder with fewer than 250 cases reported in the literature.
• Is known by a variety of pseudonyms (Table 1).
• Usually affects the long bones but can affect the axial skeleton.
• Involvement of the axial skeleton results in much of the morbidity and mortality related to this rare disorder.
• Involvement of the skull is reported but involvement of the temporal bones is extremely rare.

Table 1. Eponyms for Gorham-Stout Disease

<table>
<thead>
<tr>
<th>Eponym</th>
<th>Description</th>
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<tbody>
<tr>
<td>Gorham’s Syndrome</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
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<tr>
<td>Gorham-Stout Syndrome</td>
<td>A variant of Gorham’s Syndrome affecting the temporal bone</td>
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<tr>
<td>Morbus Gorham-Stout Disease</td>
<td>A variant of Gorham’s Syndrome affecting the temporal bone</td>
</tr>
<tr>
<td>Massive Osteolysis</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
</tr>
<tr>
<td>Idiopathic Massive Osteolysis</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
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<tr>
<td>Progressive Massive Osteolysis</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
</tr>
<tr>
<td>Massive Gorham Osteolysis</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
</tr>
<tr>
<td>Disappearing Bone Disease</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
</tr>
<tr>
<td>Vanishing Bone Disease</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
</tr>
<tr>
<td>Phantom Bone Disease</td>
<td>Disease affecting the skull and spine, especially the mandible</td>
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Case Report

A 29-year-old woman presented with complaints of aural fullness. She also stated that when she tapped on her occipital skull she experienced this fullness.

She had been diagnosed with Gorham-Stout disease at the age of 9. She subsequently required fusion of C1-C5 for cervical spine instability due to her disease.

She had also bilateral cervical lymphatic malformations. These malformations had been increasing enlarging and caused both airway and esophageal compression. She required tracheostomy to secure her airway.

Radiographically, she demonstrated mandibular involvement and extensive skull base involvement.

There was demineralization and moth-eaten changes of the osseous structures of the skull base and posterior fossa, including the dural sinuses, dems, bilateral occipital condyles, the left mastoid air cells extending to the left jugular foramen and left hypoglossal canal and left stylo mastoid process.

Figure 1. Bone windowed CT of the temporal bone. The cortex of the mastoid bone is eroded (a) and there is soft tissue opacification within some of the air cells. The occipital bone is completely destroyed from the mastoid to the occiput (b). The jugular bulb and hypoglossal canal are also altered by this permeative bony destruction (c).

Figure 2. Bone windowed CT of the temporal bone. There is destruction of the mastoid cortex abutting the inferior aspect of the meatus of the external auditory canal (a). Again, the occipital bone is completely replaced by non-calcified tissue (b). The petrous apex including the carotid canal and Eustachian tube are involved with abnormal appearing bone (c).

Figure 3. Bone windowed CT of the temporal bones. There are extensive bony changes throughout the left skull base. The left mastoid tip is completely eroded (a) and there is permeative changes extending to the jugular foramen (b). The hardware for the patient’s cerebral fusion can be seen posteriorly.

Figure 4. Hematoxylin and Eosin staining of bone taken from parietal bone. Note the dilated lymphatic (a) and vascular (b) channels as well as the dense fibrous tissue replacing the normal bony architecture. (From V. Parihar et al. Gorham’s disease involving the left parietal bone: a case report. Cases J. 2008; 1: 258.)

Discussion

Pathophysiology

• Gorham’s disease results in bone resorption and replacement with fibrovascular tissue with unusually wide capillary-like vessels (Figure 4).
• Recent Immunohistochemical analysis demonstrates high levels of a lymphatic endothelial marker (LYVE-1).
• How these abnormal vascular/lymphatic channels result in the massive bony destruction remains to be elucidated.
• Studies have demonstrated elevated humoral agents at the sites of bone and fibrovascular interface.
• The origin of these elevated enzymes remains unclear.
• The role of osteoclasts remains controversial.

Clinical Manifestations

• Head and neck manifestations of the disease include involvement of the flat bones of the skull, the mandible and the cervical spine.
• Involvement of the temporal bones has been reported in one case.
• A young child developed profound sensorineural hearing loss as her disease progressed.
• The patient in this case presented with peripheral temporal bone involvement after 20 years of active disease.
• Since she had extensive involvement of her cervical spine it is possible that the temporal bone disease represents extension of her cervical disease rather than a new focus of disease.
• Involvement of the axial skeleton is the most common cause of mortality in these patients.
• The fibrovascular/lymphovascular proliferation associated with bony destruction leads to direct involvement of the thoracic duct resulting in chylothorax which is frequently fatal.
• Involvement of the cervical spine may result in swan neck deformity if not corrected by cervical fusion.

Treatment

• There is no standard treatment.
• Active or symptomatic lesions are frequently treated with radiation.
  - There is no standard dose but many receive between 30-50 Gy per lesion.
  - The major risk is radiation induced malignancy, although this has not been reported in Gorham’s disease patients.
  - Surgical intervention is warranted for diagnostic biopsies, spine stabilization, treatment of chylothorax and bone grafting for long bone lesions.
  - There is a small series of patients who have been treated with bisphosphonates to stabilize bone lesions.
  - Some cases have been reported where alpha interferon was successfully used to treat chylothorax in combination with other treatments.
• Alpha interferon has anti-angiogenesis properties.

Summary

• Gorham’s disease is a rare entity of unclear etiology that typically presents early in life.
• Manifestations in the head and neck are fairly common; however, direct involvement of the temporal bone is very unusual.
• In the long bones, resection and grafting is effective.
• Involvement of the axial skeleton results in much of the morbidity and mortality associated with the disease.
• In the axial skeleton, radiation and medical therapy may halt the progression of disease.

References