Gorham Disease


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Author Affiliation | Disclosures

Authors’ Disclosure Statement: The authors report no actual or potential conflict of interest in relation to this article.

Take-Home Points

- Gorham disease is a rare condition that manifests as an acute, spontaneous osteolysis.
- There is no clear hereditary pattern of transmission. Bones of any type or location can be affected.
- Imaging studies are nonspecific, but show permeative osteolysis involving the subcortical and intramedullary regions and typically affect regional, contiguous bones, without adjacent sclerosis, somewhat resembling osteoporosis.
- Tissue biopsy is indicated to rule out other potential etiologies of osteolysis, and the histologic findings help confirm a diagnosis of Gorham disease.
- There is no single or combined treatment modality that is considered as the gold standard. Surgical treatment includes resection of the lesion and reconstruction. Also, antiosteoclastic medication can be used.

Gorham disease, a rare condition of unknown etiology, manifests as acute, spontaneous osteolysis associated with benign hemangiomatosis or lymphangiomatosis, which presents as skeletal lucency on radiographs, prompting the classic eponym of vanishing bone disease.\textsuperscript{1-6} There is no evidence supporting the idea that osteoclasts are present in any meaningful amount in the resorption areas or that local reparative osteogenesis occurs.\textsuperscript{4,6}

Jackson and colleagues first described idiopathic osteolysis in 1838,\textsuperscript{1,2} and Gorham and Stout\textsuperscript{3} introduced the syndrome to the orthopedic community in 1955. Since then, few strides have been made in identifying the disease origin.\textsuperscript{1,2,4} Diagnosis is possible only after meticulous work-up has excluded neoplastic and infectious etiologies.\textsuperscript{7,8}

Clinical Presentation

Gorham disease affects patients ranging widely in age, from 2 months to 78 years, but typically presents in those under 40 years. There is a questionable predilection for males but no correlation with ethnicity or geographic region. There is no clear hereditary pattern of transmission.\textsuperscript{7} Although the bones of the head, neck, and upper extremities are involved in most cases, bone of any type or location can be affected.\textsuperscript{6} Pelvic bones seem to be involved least often.\textsuperscript{6,7}

Initial clinical presentation varies considerably but typically involves prolonged soreness in the affected region and, rarely, acute pathologic fracture.\textsuperscript{1,2,4} The nonspecific nature of complaints, lack of markers of systemic illness, and rarity of the disease contribute to delayed diagnosis.\textsuperscript{1,2}
Imaging

Plain radiographs show permeative osteolysis involving the subcortical and intramedullary regions and typically affecting regional, contiguous bones, without adjacent sclerosis, and somewhat resembling heterogeneous osteoporosis.

Computed tomography (CT) better defines the severity and extent of these changes. Progression can result in osseous tapering, or “pointing” at lytic margins, forming cone-shaped spicules. In progressive cases, there is an “extraosseous” stage characterized by frank cortical destruction and true “disappearance” of bone, with extensive soft-tissue edema.
Figure 2.
(Figures 2A, 2B).

Magnetic resonance imaging shows an infiltrative and irregular T2 hyperintense signal throughout regions of bone affected by osteolysis, but this finding is not characteristic. There is heterogeneous enhancement on postcontrast sequences, and, though masslike enhancement is absent, signal abnormalities may extend into adjacent soft tissues. These changes indicate inflammation and hemorrhage of various degrees interspersed with scant fibrous tissue.\textsuperscript{8-10}

Figure 3.
(Figures 3A, 3B).

Bone scintigraphy using technetium-99m is similarly nonspecific, typically revealing radiotracer uptake that is consistent with bony reaction to an underlying osteolytic process.

Figure 4.
(Figure 4) but turning negative with ongoing resorption. In some cases of Gorham disease, bone scintigraphy did
not reveal a significant increase in activity, such as would be expected in a vascular malformation or purely angiomatous neoplasm. Similar findings could be attributed to a variety of pathologies, including primary bone tumor, metastasis, or even osteomyelitis.\textsuperscript{8,10}

Positron emission tomography/CT typically shows foci of increased metabolic activity in the areas of osteolysis.\textsuperscript{10}

**Diagnosis**

There have been 8 histologic and clinical criteria described to diagnose Gorham disease: (1) biopsy positive for presence of angiomatous tissue, (2) complete absence of any cellular atypia, (3) lack of osteoclastic response and lack of dystrophic calcifications, (4) evidence of progressive resorption of native bone, (5) no evidence of expansive or ulcerative lesion, (6) lack of visceral involvement, (7) osteolytic radiographic pattern, and (8) no concrete diagnosis after hereditary, metabolic, neoplastic, immunologic, and infectious work-up.\textsuperscript{4,6} These criteria confirm that the diagnosis can be rendered only after exclusion of neoplastic and infectious etiologies through clinical and laboratory work-up, imaging studies, and tissue sampling.

Tissue biopsy is indicated to rule out other potential etiologies of osteolysis, and the histologic findings help confirm a diagnosis of Gorham disease. Biopsies typically show a progressive osteolysis that is consistently associated with a benign but abnormal vascular proliferation that in many cases has characteristics of lymphatic endothelium. The apparent bony destruction is largely attributed to this process.

![Figure 5.](ajo04606458e_f5.jpg)

**Figure 5.**

**(Figures 5A-5D).**\textsuperscript{11,12}

The differential diagnosis includes infection (osteomyelitis, Brodie abscess), benign tumors (eosinophilic granuloma/Langerhans cell histiocytosis), malignant tumors (Ewing sarcoma and angiosarcoma), inflammatory conditions (eg, apatite-associated destructive arthritis), endocrine disorders (eg, osteolytic hyperparathyroidism), benign non-neoplastic conditions (venous or venolymphatic malformation), and other syndromes that present with osteolysis.\textsuperscript{1,2} Nevertheless, progressive and unusually substantial bone destruction without evidence of repair is almost pathognomonic for Gorham disease.\textsuperscript{9}
Treatment

Table.
Although no single or combined treatment modality is considered the gold standard (Table)\(^1\), management of Gorham disease generally centers on radiation therapy for local control of large and painful lesions and on surgical intervention for pathologic progression that would otherwise result in substantial functional limitations.\(^2\) Also described for treatment are antosteoclastic medications (bisphosphonates), which are often used in conjunction with radiation and/or surgical intervention.\(^2,4\) The newer literature cites some benefit of using various experimental modalities, including a combination of interferon alfa-2b and low-molecular-weight heparin,\(^13\) and even propranolol.\(^14\)

Surgical treatment usually includes lesion resection and subsequent reconstruction using combinations of bone grafts (allogenic) and prostheses. Bone graft alone is quickly resorbed and has not been found to be beneficial.\(^1,2,4,20\)

**Key Info**

**Figures/Tables**

**References**

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Nov 2018  
The *American Journal of Orthopedics* ©


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