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Gorham disease of the mandible: Radiographic findings and radiotherapy response

Abstract

Gorham disease, a rare condition of unknown etiology, is characterized histologically by bone disintegration and endothelial proliferation. We describe serial imaging findings, the treatment course, and radiotherapy response in a patient with pathologically confirmed Gorham disease involving the right mandible. Progressive mandibular resorption was managed initially with multiple surgical resections and reconstruction, followed by external-beam radiotherapy, which was an effective treatment method in this case. The patient's reconstructed mandible is functional and he is asymptomatic. Our literature review provides further insights regarding the clinical, radiologic, and pathologic behavior of this entity and examines the available treatment strategies.

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Introduction

Gorham disease (also referred to as *massive osteolysis*, *vanishing bone disease*, or *disappearing bone disease*) is an uncommon, progressive condition in which bone is resorbed and replaced by proliferation of endothelial and fibrous tissue. It can occur in any bone.¹ Histologic findings are often characterized by thin-walled vascular channels in bone, although some authors have been unable to identify definite pathognomonic features.^{2,3} Etiology remains unknown, and because of the paucity of reported cases in the literature, little has been established about the disease process since the condition was first described by Jackson in 1838.⁴

This disease has a progressive course in most patients, although some reports describe self-limiting cases.⁵ Current treatment methods include surgery (effective when all involved tissue can be excised) and radiation therapy. In general, the disease is characterized by aggressive resorption of local bone. However, extension into adjacent soft tissues has also been reported.^{6,7}

In this report, we describe the unique presentation, radiologic findings, and treatment course in a patient with pathologically proven Gorham disease of the mandible.

Case report

A 34-year-old man with no significant medical history presented with initial symptoms of trismus and jaw misalignment. His primary care provider referred him to a local maxillofacial surgeon (G.E.A.) for further evaluation and treatment.

Plain-film x-rays revealed a malformed right mandibular condyle with a subtle adjacent mandibular ramus fracture (**figure 1**). The left condyle and mandible were normal. Three months later (during which the patient experienced trauma to his right jaw at work), repeat panoramic x-rays revealed no improvement. Because of absence of pain or noticeable symptoms, he opted for observation rather than surgery and was followed with serial imaging only.

Initial panoramic x-ray shows subtle loss of the lateral border of the right mandibular ramus (solid arrow) compared with normal density of the left mandibular ramus (open arrow).



One year after the initial radiographic abnormalities were noted, repeat panoramic x-rays of the mandible revealed progressive bone loss around the tooth roots (**figure 2**). The patient reported experiencing increasing frequency and intensity of dull pain in the right mandibular ramus during the period since he had last been seen. The maxillofacial surgeon performed resection and stabilization of the right ascending ramus of the mandible and the condylar area (the site of

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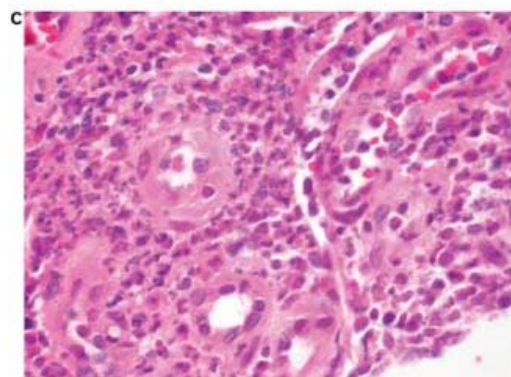
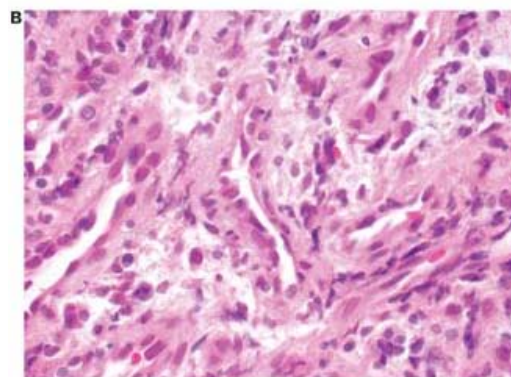
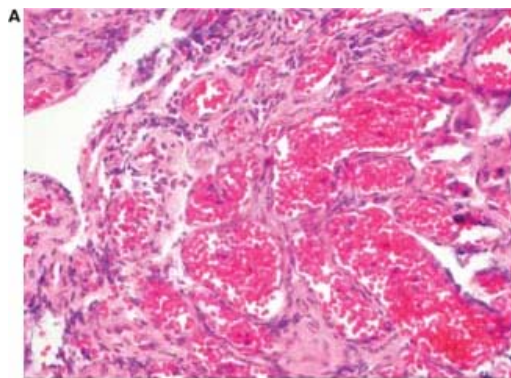
significant bone resorption) and mandibular reconstruction with a synthetic condylar bone plate. No evidence of malignant tumor or infection was found on pathologic evaluation, and surgical notes indicated "bone fragment with mild chronic inflammation and hemorrhage consistent with fracture." Follow-up with serial x-rays continued.

Approximately 1 year after the initial examination, panoramic x-ray shows significant loss of osseous cortices of the entire ramus of the mandible (arrows), including complete absence of the condylar neck and head and mandibular coronoid process.



Fifteen months after the initial evaluation, x-rays revealed further mandibular bone loss progressing toward the midline. The maxillofacial surgeon performed further resection and mandibular reconstruction at that time, and the pathology specimen revealed heavy proliferation of vascular channels with massive osteolysis (**figure 3**).

Micrographs reveal (A) closely spaced, blood-engorged vascular channels lined by cytologically benign endothelial cells (original magnification $\times 200$), (B) closely spaced small vessels with minimal intraluminal blood, suggesting lymphangiomatous differentiation (original magnification $\times 400$), and (C) closely spaced small vessels with moderately intense acute and chronic inflammatory cell infiltrate (original magnification $\times 400$)



Histologic examination of the resected material showed dense proliferation of contiguous and focally anastomosing vascular structures, most of which were engorged with blood. No nuclear atypia, tissue necrosis, or increased mitotic activity was seen. Focally, the minimal amount of intervascular connective tissue exhibited a mixed inflammatory cell infiltrate (consisting of neutrophils, lymphocytes, histiocytes, and a few plasma cells). The microscopically examined tissue contained no bone elements (osteoclasts, osteoblasts, or osteoid). The histologic preparations were sent for outside pathologic consultation, and the diagnosis of Gorham disease was confirmed.

The patient was referred to the Department of Radiation Oncology at Huntsman Cancer Hospital. Postoperative radiation therapy was recommended because of continued disease progression, and the patient consented to external-beam radiation. Unfortunately, he did not return for his scheduled radiation-planning session, and repeated attempts to contact him were unsuccessful. However, 8 months later he returned to radiation oncology with further mandibular bone resorption, which required additional mandibular resection and hardware reconstruction. Again, the patient consented to postoperative external-beam irradiation, and he completed the recommended course.

On the basis of our review of the literature, radiotherapy targeted the mandibular resection bed and mandibular remnant. External-beam radiation was administered at 2 Gy/day for 20 fractions (25 elapsed days) to a planned total dose of 40 Gy with opposed lateral fields.

Since completion of radiation therapy, there has been no clinical or radiographic evidence of further bone resorption. The patient has experienced mild trismus, but his reconstructed mandible has remained functional and he is otherwise asymptomatic.

Discussion

Many facets of Gorham disease remain a mystery because fewer than 100 cases have been described in the literature. In the 1950s, Gorham et al presented information on the first 24 cases and an overview of the disease-process manifestations, and they described the pathologic-specimen findings in 8 cases.^{8,9}

The causative factors for Gorham disease are unknown. There is no evidence of an infectious or

other disease etiology, although some investigators consider it a neoplastic entity or propose that it falls within a category of idiopathic osteolytic syndromes.^{1,5,7,10,11} One theory is that bone loss is secondary to the pressure effect of a soft-tissue mass. According to Dunbar et al, the condition is characterized by localized proliferation of thin-walled endothelial-lined capillaries or sinusoidal channels (osteolytic angioma), which results in destruction and resorption of bone.⁵ The shoulder girdle and pelvic bones are the most commonly reported sites of disease although, theoretically, any bone could be involved.

Diagnosis also can be unclear. Because of this difficulty, Heffez et al have suggested specific criteria to distinguish Gorham disease from other entities involving bony destruction.¹² Their criteria for Gorham disease are:

- Positive biopsy results for angiomatous tissue;
- Presence of an osteolytic radiographic pattern;
- Evidence of local progressive osseous resorption;
- Minimal or no ectoblastic response;
- Absence of cellular atypia, dystrophic calcification, visceral involvement, or an expansile or ulcerative lesion;
- Absence of a hereditary pattern; and
- Absence of a metabolic, neoplastic, immunologic, or infectious cause.

Radiologic findings of Gorham disease are not widely available. Differential diagnoses for an aggressive osteolytic lesion on plain-film x-rays include osteolytic metastatic foci; destructive lesions such as a rhabdomyoma, teratoma, and eosinophilic granuloma; and neuroblastoma in a child (all of which would be expected to show a more aggressive clinical presentation than seen in Gorham disease). An early plain-film finding is osseous loss of density, which has been described as similar to the loss of density in osteoporosis.^{3,10} A commonly reported finding is continued loss of normal-appearing osseous structures, sometimes with extension into the surrounding osseous anatomic landscape and even across joints to contiguous bones. Bone loss can continue over a period of years, then stabilize.^{3,10,13}

Computed tomography (CT) shows osseous resorption and osteolysis. Noncontrasted bone algorithm images, with complete evaluation of the surrounding osseous structures, are most helpful.^{3,10} Magnetic resonance imaging (MRI) can also demonstrate hypointensity to isointensity on T1-weighted images and hyperintensity on T2-weighted images. Precontrast T1-weighted images may be most helpful for lesion evaluation, as they are for extracranial head and neck lesions, because of their excellent contrast differentiation between a mass and normal extracranial fat. Lesions with increased vascularity typically show heterogeneous enhancement on both CT and MRI postcontrast images, but MRI's fat-saturation techniques may greatly assist in evaluating the full extent of the lesion.¹³

Pathologically, Gorham disease is characterized by proliferation of hemangiomas, and often lymphangiomatous-like, tissue consisting of freely anastomosing, thin-walled vascular structures lined by cytologically bland endothelial cells.^{8,14,15} In some ways, this vasoformative process is histologically similar to skeletal angiomas, but it is more destructive.

Massive osteolysis is usually monocentric, whereas skeletal angiomas is more often multicentric.^{5,6,16} As vascular proliferation of massive osteolysis progresses, whole bones or several adjacent bones are completely resorbed. Often, periosteal fibrous tissue is destroyed or reduced to only a thin band. In spite of massive bone loss, osteoclastic activity is minimal or absent. Occasionally, the proliferating vascular mass extends into periosteal soft tissue. Grossly, the bone is soft and spongy. Over time, vascular tissue is replaced by fibrous tissue, with complete absence of bony remnants.^{5,7,10}

In contrast, skeletal angiomas is characterized by incomplete bone resorption. Periosteum and a thin shell of reactive new bone, with prominent osteoblastic activity, remain at the periphery of the lesion, and there is little or no replacement of the vascular tissue by fibrous tissue.

No standard therapy for Gorham disease has been established, again because of its rarity.⁵ Surgery and radiation therapy are commonly described, and both have been successful in controlling disease in select cases.^{2,5,17} As illustrated in this case, early intervention with definitive radiation therapy in moderate doses, from 30 to 45 Gy in 2-Gy fractions, appears to result in an excellent patient outcome. In some reports, radiotherapy has led to recalcification.^{5,17} However, substantial bone deformity may be the final result.⁵

The prognosis for patients with Gorham disease is generally good unless soft tissue or vital structures are involved. Some patients will have a favorable outcome regardless of treatment method, and in a few, spontaneous regression has been reported.¹⁸

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