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# Vanishing Bone Disease of Mandible: A Diagnostic Challenge

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## Abstract

**ABSTRACT**: Gorham's disease is a rare entity involving clinical and radiological disappearance of bone by proliferation of non-neoplastic vascular tissue(1). It was first reported by **Jackson** in 1838 in the arm. Later on it was further elaborated **by Gorham and stout** in 1955, and hence the name of the disease. It can affect one or multiple bones in the patient with very less prevalence in the head and neck region.

**Keywords:** Gorham's disease; osteolysis; vanishing bone disease.

## Introduction

Gorham's disease or vanishing bone disease is a disease with a very vague understanding of the etiology. With a course of progressive bone loss, this idiopathic massive osteolysis is seldom found in the head and neck region(2) Though any bone can be affected, the predominantly affected bones include the pelvis, humerus, axial skeleton(3). The maxillofacial involvement is less, compared to the other areas. The mandible is the most commonly affected bone in the maxillofacial region, with only upto 50 cases reported till date(4). **Escande et al**. (5) reported that only 41 cases of this syndrome involved the head and neck, with only five cases involving paediatric patients and maxillofacial region. The lesion initially has sluggishly flowing capillary like vessels, which results in a drop in the pH in addition to creating a hypoxic environment locally, thus favouring bone resorption by the production of various hydrolytic enzymes(6).

Here, we present 3 cases of this massive and rapid osteolytic disease of mandible, and also a literature review of the treatment options for this rare disease.

## Case Presentation

**Case 1**: A male patient aged 52 years, reported to the department of dentistry, AIIMS Patna, with a chief complaint of pain and swelling in his right lower back jaw

region since one and half years. The medical history was non-contributory. His habits included tobacco chewing for the past 15 years in a frequency of 5-6 times/day. On extraoral examination, the patient had a large depression over the right body of mandible causing a gross facial asymmetry and a mild mandibular deviation to the left side. Extraoral sinus opening was present in relation to the right para symphysis region. On intraoral examination, there was bone exposure in the region of the lower anteriors, with loss of the lower anterior teeth. The OPG & CT face showed resorption of the right mandibular body, the right ramus, and the right coronoid process with only a residual rim of basal bone extending from the region of 41-48. The residual bone did not show any evidence of periosteal reaction and appeared to have ragged borders [ fig 1&2].

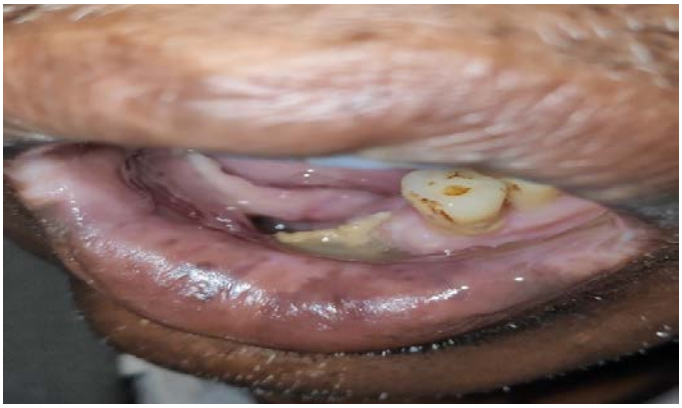

Fig 1

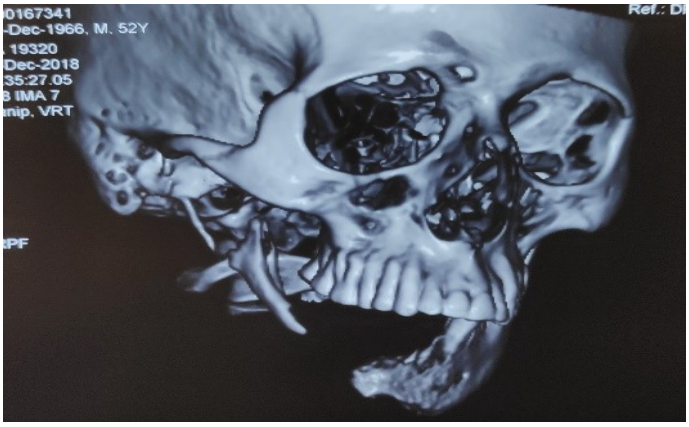


Fig 2

**Case 2**: A male patient aged 50 years, reported to the department of dentistry, AIIMS Patna with chief complaint of pain & bilateral swelling of the lower back jaw region since1 year. There was no relevant medical history. On examination there was an extra oral sinus opening over the bilateral body of mandible region accompanied by depression. There was gross facial asymmetry. The mouth opening was restricted to about 10 mm. The CT scan revealed gross bone resorption of both sides of the mandible with interspersed bony fragments [fig 3&4].

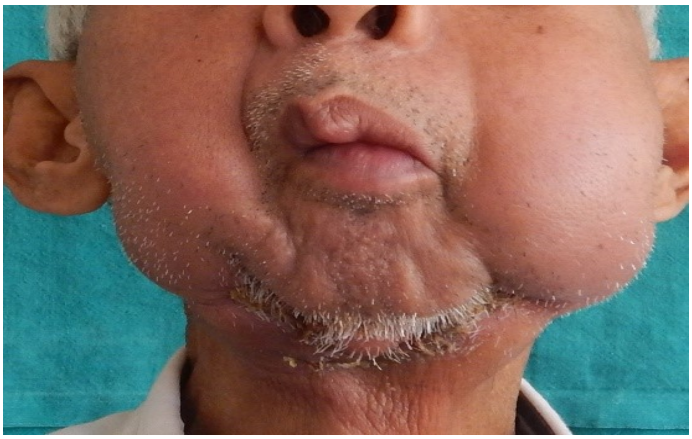

Fig 3

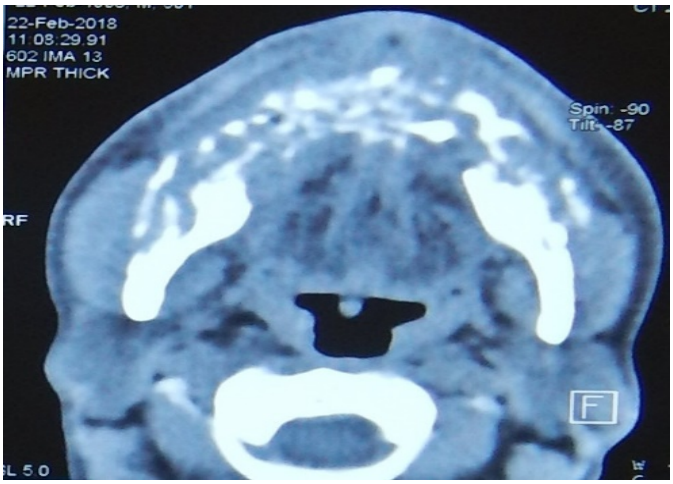


Fig 4

**Case 3:** A male patient aged 60 years, reported to the department of dentistry, AIIMS Patna, with a chief complaint of pain and swelling in his left lower back jaw region since one and half years. There was no relevant

medical history. Extraoral sinus opening was present in relation to the left body of mandible region. On intraoral examination, there was bone exposure in the left molar region with loss molar tooth. The CT scan revealed extensive bone destruction on left body of mandible of size 3.6*2.8 cm approx. [fig 5&6].

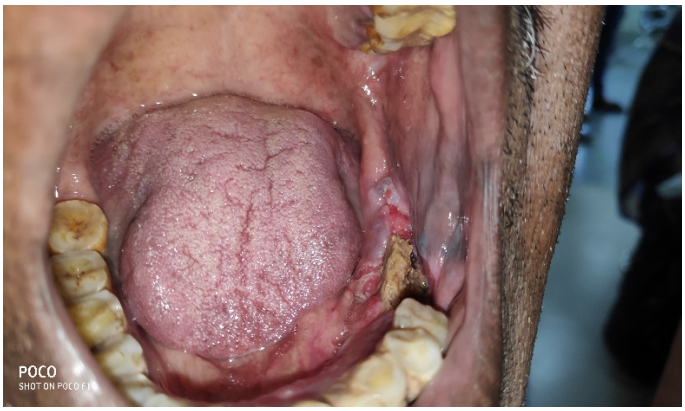


Fig 5

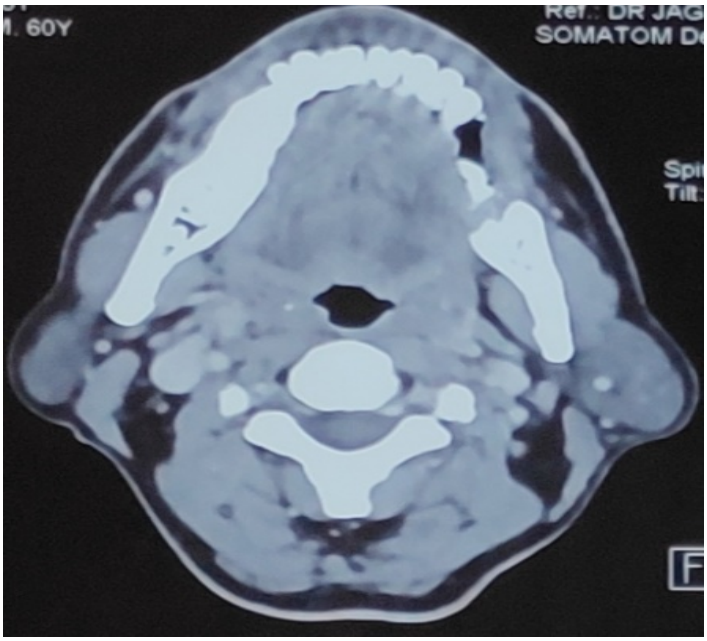


Fig 6

**Histopathology**

Histopathology of all 3 the cases were similar**.** Biopsy of the first case initially done at a private centre showed hyperplastic stratified squamous epithelium with pseudo-epitheliomatous hyperplasia and marked chronic inflammatory cell infiltrate at dermo-epidermal junction.

A repeat biopsy of the first case was done at the our department, which stressed the evidence of an acute or chronic inflammatory pathology with no features of cellular atypia and was suggestive of an angiomatous lesion Biopsy of the case 2 &3, also showed similar findings.

All blood investigations (CBC, LFT, KFT, PTH) were within normal range and chest x-ray was showing normal radiographical features. Differential diagnosis of osteomyelitis, Rheumatoid arthritis, central giant cell granuloma; secondaries of jaws, osteogenic sarcoma; Histocytosis, Stills disease, endocrine disorders like hyperparathyroidism and other osteolytic lesions of jaws, which needed to be excluded. On the basis of excluding criteria for other osteolytic lesion diagnosis of Gorham's disease was made (**Heffez et al**). Patient was managed with surgical resection with a clear margin and reconstruction of resultant defect with reconstruction plate (case 1) and loco-regional flap (case 2&3) and were send for radiation therapy. Postoperatively, patients were asymptomatic and on under regular follow up.

**Discussion**

This is a queer phenomenon involving the slow resorption of bone(7). To this day, the exact etiology and trait of the disease process is a mystery. It is a disease that occurs majorly in young adults with no specific sex predilection(8), but **Heffez et al**(9) reported a specific age group of 1 month to 66 years. The pathophysiology of the disease process has been described by many authors. **Pazzaglia et al**(10) reported that disease occurs in 2 phases, with active bone resorption occurring in the first phase of the disease, accompanied by pain and soft tissue swelling. The second phase is dormant. **Perschbacher et al (11)** described this lesion as a non-malignant, cancer-like proliferation of haemangiomatous or lymph angiomatous tissue. Localised vascular expansion leading

to the derangement of the osteoblast to osteoclast balance and increased hyperaemia is postulated to be the causative factor behind this massive osteolysis, as reported by **Gorham et al**(7). **Devlin et al** (12) found out the comparable levels of IL-6 in those with the disease, in whom the level was 7 times the normal limit. He also reported a case in which there was massive resorption of the mandible which extended to involve the maxilla, zygoma, right parietal region and cranium(12). Overall, Gorham's is a disease that easily affects the abutting bones and has no regard for joint boundaries(13). **Dickson et al (14)** reported the first cytochemical investigation of the disease. They were of the opinion that mononuclear phagocytes, multinuclear osteoclasts, and vascular endothelium participate in the resorption process.The histopathological scenario of this condition as reported by **Korsiç et al (15),** involves the proliferation of endothelium lined thin walled capillaries. Though, generally the prognosis of this condition is good,5 fatal cases with severe chest involvement have been reported(13). Routine lab analysis is within normal limits, though some cases with pathologic fracture have reported an increase in alkaline phosphatase levels(9).

**Heffez et al** (9) suggested 8 criteria for definitive diagnosis of Gorham's:

1. Angiomatous tissue presence in biopsy
2. No cellular atypia
3. Little or no osteoclastic response and no dystrophic calcifications
4. Evidence of progressive resorption of local bone
5. Non-expansive, non-ulcerative lesion
6. Negative visceral involvement
7. Osteolytic radiographic pattern
8. Non-contributory hereditary, metabolic, neoplastic, immunologic, and infectious etiology

**Treatment options:** From 2010, upto the present, various treatment modalities have been proposed, some of them being sodium fluoride, vitamin D, calcium, alpha interferon(16), calcitonin and reconstructive surgery with a micro-vascularized graft(17). These bone grafts have been reported with varying success rates depending on the activeness of the disease. Daily subcutaneous calcitonin injections and infusions of pamidronate every 3-4 months were some of the older treatment policies(12). **Bouloux et al** (18) tried a combination of Sulfamethoxazole (400 mg) and trimethoprim (80 mg) supplemented with phenoxy-methyl penicillin (500 mg) for 4 months in a 19 years old female, but no follow up has been described(18).

Radiotherapy has had a decent success rate. Due to the radiosensitive nature of endothelial cells, **Ricalde et al (19)** achieved positive results with a dosage of 45 Gy, with the average dosage being 30.6 Gy. Stoppage of the osteolytic process along with recalcification has been reported in certain studies(17,20,21).

Another treatment modality which has been used is interferon alpha-2b, an anti-angiogenic agent which is believed to cease the proliferation of blood vessels and also the level of IL-6(16,22). Surgical resection of the affected site along with osseous graft placement has not been much of a success due to resorption of the graft(11), though surgical excision by **Dong et al**(23) resulted in stoppage of further disease progression. Bisphosphonates and calcitonin have been used predominantly for the stoppage of local osteolysis(24).Treatment with zolendronic acid has also been documented. **Kuriyama et al**(25) reported a case of gorham's with pleural effusion that was treated with zolendronic acid and peg-interferon alpha 2b successfully. **Avelar et al**(26) reported maintenance of bone volume with the usage of 4 mg zolendronic acid for 30 minutes, once a month), calcium (500 mg), and vitamin D (400 IU) administrated once a

day for 12 months. RANK-Ligand inhibitor Denosumab, was used by **Liu et al**, in a dosage of 60 mg administered as a single cutaneous injection every 6 months, as it was believed to inhibit the development of osteoclasts(24). Applying compression with bandages, manually massaging to increase the lymphatic clearance and also for protecting skin and soft tissue from inflammation has shown to be beneficial(27).

**Conclusion**

Gorham disease is an extremely rare disease with a multifactorial etiology. Diagnosis is mainly based on clinical presentation, radiologic imaging , and biochemical analysis and histo-pathologic findings. We should also exclude the possibilities of neoplastic, inflammatory, neuropathic, and metabolic disorders showing osteolytic nature. Even though there are a multiple treatment modalities option available but the ideal management strategy for this is still a challenging one and it requires a lot of future research.

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