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Case Report

Gorham's disease of the maxilla – A rare case report with literature overview

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ABSTRACT

Gorham's disease is a rare condition characterized by progressive osteolysis of bone ultimately resulting in the total disappearance of bone. The etiology is unknown and the disease predominantly affects the pelvis, humerus, and axial skeleton. Because of its unusual, ambiguous presentation and rare occurrence, the disorder goes unrecognized and is often masqueraded by other disorders. The diagnosis of this disorder is by exclusion. About 50 cases of Gorham's disease involving the maxillofacial region are reported to date and most of them involve the mandible. Exclusive involvement of maxilla is documented in only 4 cases. We report an exceptional case of Gorham's disease of the maxilla in a 68-year-old male patient presenting with chronic pain and masquerading as an odontogenic infection.

Introduction

Gorham's disease is a rare destructive condition of the skeletal system resulting in progressive resorption and disappearance of osseous structure with subsequent replacement by vascular and fibrous connective [1,2]. Various synonyms used in the literature to describe the condition include phantom bone, disappearing or vanishing bone disease, acute spontaneous absorption of bone, hemangiomatosis, lymphangiomatosis, idiopathic osteolysis and Gorham's disease [3]. Jackson in the year 1838, was the first to report a case of disappearing humerus that led him to title his work as "a boneless arm" [4]. In 1924, first case was reported in the jaws by Romes. Complete lysis of mandible was reported by Thoma in 1933 [5]. This disease also known as the Gorham-Stout disease, named after two physicians who presented a case series of 24 patients in 1954 and 1955 [6,7].

The condition can manifest in any part of skeleton, but commonly involves the skull, shoulder and pelvic girdle [8]. It is commonly detected after fracture or injury and pain may be the presenting sign of the disease. In the maxillofacial region most common symptoms are pain, loose teeth and pathological fractures [9]. In the maxillofacial region mandible is frequently involved alone or with maxilla but exclusive maxillary involvement is reported only in 4 cases. We describe a case presenting with chronic pain in the upper front teeth and the right maxilla masquerading as an odontogenic infection and was subsequently diagnosed as Gorham's disease on the basis of radiographic, laboratory and histopathological findings.

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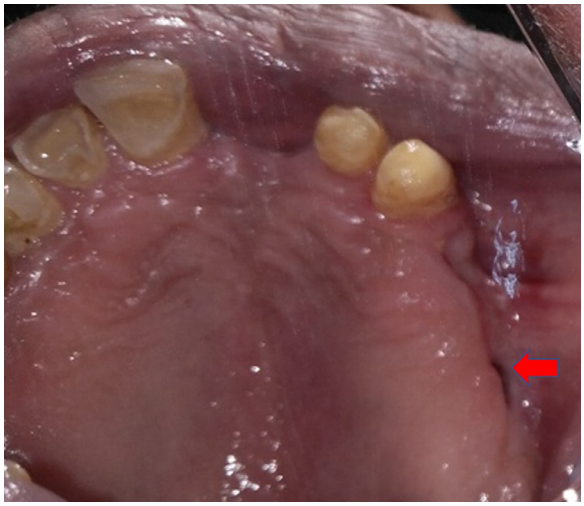

Fig. 1 – Intra-oral view depicting the presence of edentulous region and mucosal fistula.

Case report

A 60-year-old male patient reported to the department of Oral Medicine and Radiology with a chief complaint of pain and swelling on right side of face and upper front teeth for 3 months. Patient was apparently alright 3-4 months back when he got his upper right premolars and molars extracted due to mobility. Eventually he developed severe pain, associated with burning sensation on the right side of the face. Pain increased in severity in the last 15-20 days which radiated to the right eye and right side of nose. Pain was accompanied by heaviness on the right side of face. His medical history was insignificant and non-contributory.

Extra oral examination revealed mild diffuse swelling on the upper right side of face extending anteroposteriorly from right ala of nose to the right zygoma. The swelling was afebrile, tender and soft in consistency. The overlying skin was normal. Hypoesthesia was elicited on the right maxillary region. Intraoral examination revealed localised, erythematous swelling of the attached and interdental gingival of 12& 13. Swelling was tender and pus discharge was present on provocation. The right upper molars and premolars were missing. Mucosal fistula was evident in the edentulous first molar and premolar region with pus discharge on provocation. (Fig. 1) Based on the clinical findings a provisional diagnosis of chronic suppurative osteomyelitis was given. Differential diagnosis included maxillary sinusitis and oroantral fistula.

Panoramic radiograph (Fig. 2) revealed bone loss of the upper right posterior alveolar ridge extending till the maxillary tuberosity and floor of the maxillary sinus. Maxillary cross-sectional Occlusal radiograph (Fig. 3) revealed an extensive ill-defined radiolucency interspersed with faint radiopaque specks giving a moth-eaten appearance extending till tuberosity and hard palate was resorbed till midline. No expansion or cortical reaction was observed. Calcifications and sequestrum formation were not evident. Contrast enhanced CT (Fig. 4) revealed lytic destruction of maxilla on right side, nasal process

Fig. 2 – Panoramic radiograph revealing bone loss in the right posterior maxilla extending to the maxillary tuberosity.

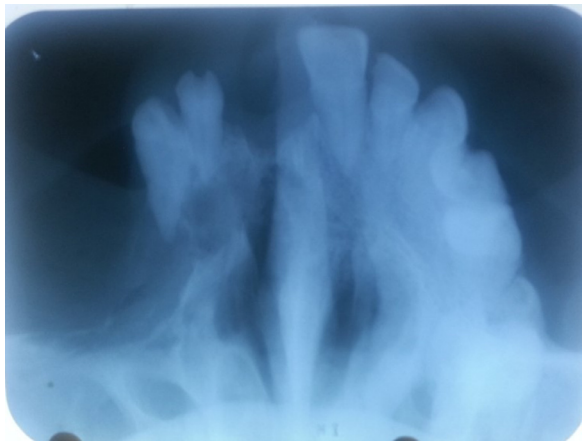

Fig. 3 – Occlusal radiograph demonstrating ill-defined radiolucency interspersed with faint radiopaque specks.

of right maxillary bone, anterior part of nasal septum, palate on right side, floor, lateral and medial wall of right maxillary sinus. Rest of the right maxillary sinus showed thick sclerosed walls suggestive of chronic maxillary sinusitis. Sub centimetre level 1 b lymph nodes were seen bilaterally.

Skeletal survey of humerus, clavicle, pelvic girdle and tibia did not reveal similar osteolytic lesions. Pus obtained was sent for culture and the results were positive for growth of a few streptococcal group of organisms. This denoted secondary bacterial infection and ruled out the possibility of deep fungal infection.

Biochemical analysis (Serum Calcium, alkaline phosphatase, Parathyroid and thyroid levels, Blood sugar levels) was performed to rule out metabolic disease. All the values were within normal range. Curettage of the affected part was undertaken and the curetted specimen was submitted for histopathological examination. Intraoperative findings suggested complete absence of bone on reflecting the mucosa with profuse bleeding. The histopathological examination of soft tissue showed increased chronic inflammatory infiltrate. No cellular atypia was observed. Histopathological examination of bone was nonspecific ruling out any malignancy or infection. After excluding other disorders like metabolic diseases, malignancy and infections a diagnosis of Gorham's disease of the maxilla was given due to presence of diffuse osteolysis of right maxilla coupled with normal histopathological and laboratory findings. Healing occurred uneventfully. The

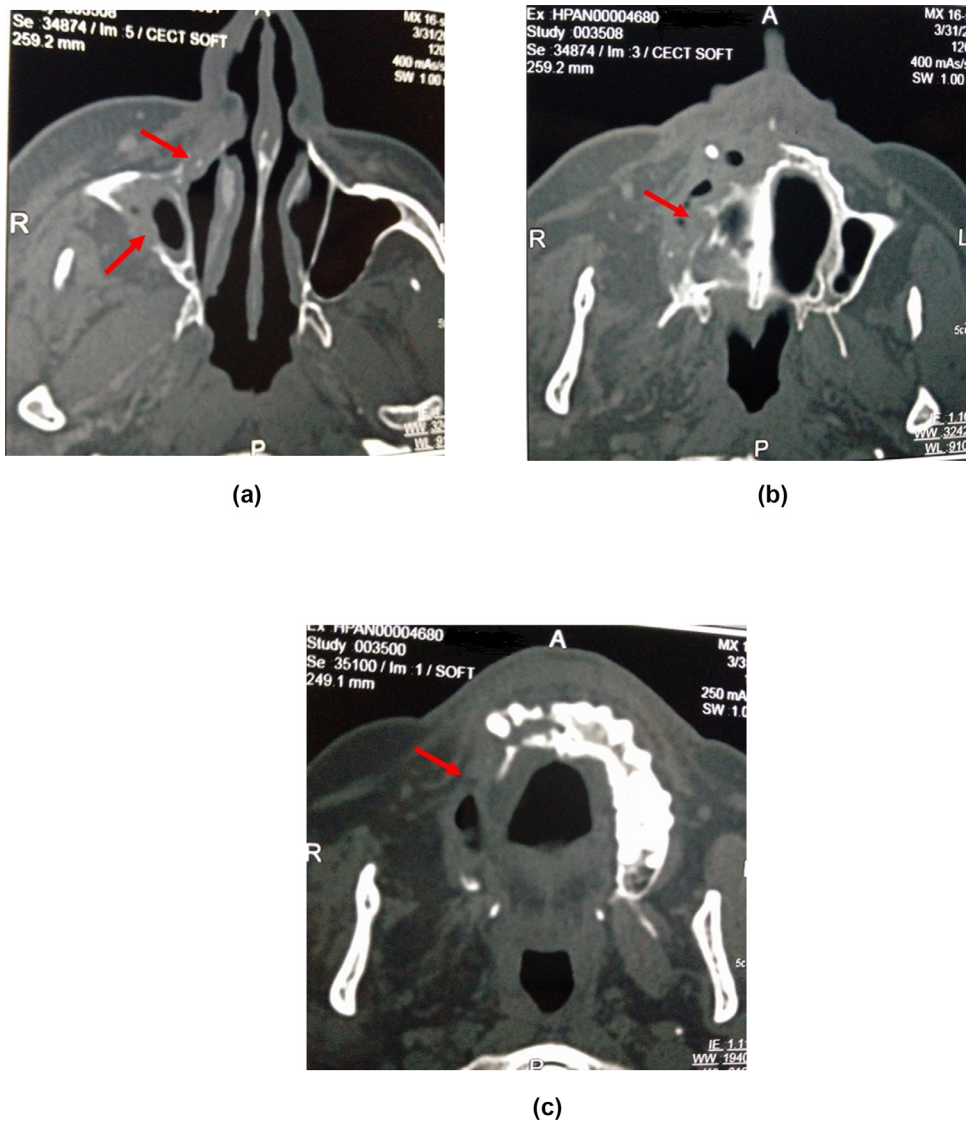

Fig. 4 – (A) Contrast-enhanced CT in axial section depicting lytic destruction of floor, lateral and medial wall of right maxillary sinus.
(B) Contrast-enhanced CT in axial section demonstrating destruction of nasal process of right maxillary bone and anterior par.t of nasal septum.
(C) Contrast enhanced-CT in axial section depicting lytic destruction of maxilla and palate on right side.

patient was rehabilitated with an acrylic obturator to cover the osteolytic defect and facilitate chewing. The intensity of pain has reduced but burning quality still persists. He was advised gabapentin 300mg twice daily for 2weeks and the patient is under follow up for 6 months and no progression of osteolytic process was observed.

Discussion

Gorham's disease is a rare occurrence and commonly manifests in pelvis, humerus and axial skeleton [5,6]. About 200 cases are described in international literature and 50 cases in the maxillofacial region [10]. Most of the previously reported cases of the maxillofacial region affect the mandible alone or with maxilla. Exclusive involvement of maxilla is rarely described in the literature. Only 4 cases are reported involving the maxilla till date and ours is the 5th case (Table 1) [11-18]. The remaining 4 cases described in Table 1 showed multicentric involvement of other craniofacial bones. The initiation of the disorder begins in the childhood and may manifest at any time between 1 month and 75 years [19]. This patient presented to us at 68 years of age but the process of mobility and extraction of his teeth had started 2 years ago. Although no sexual predilection has been found, Huvos [20] reported 60% of all cases manifesting in men and is substantiated in the present case [7]. According to Table 1 the cases reported in the maxillofacial region showed a male predominance.

Table 1 – General characteristics of cases involving the maxilla and craniofacial bones.

	Authors	Age	Gender	Site	Other bones involved
1	Rodriguez-Vazquez JR et al (2019) [11]	53	M	Left Maxilla	-
2	Zakhary I et al (2016) [12]	57	F	Right Maxilla	-
3	He J et al (2012) [13]	37	M	Left Maxilla	-
4	Perschbacher et al (2010) [14]	56	M	Left Maxilla	-
5	Sinha R et al (2017) [15]	38	M	Right Maxilla	Mandible
6	Benhalima H et al (2001) [16]			Maxilla	Mandible, orbit, cranial bones and cervical spine
7	Ohya T et al (1990) [17]	66	M	Maxilla	Mandible
8	Oujilal et al (2000) [18]	17	M	Maxilla	Ramus, sphenoid, temporal and occipital

Although 60 years have elapsed since the description of the disease, the etiology is still very speculative. There are many hypotheses of the aetiologies such as post-traumatic hyperaemia, changes in blood pH, hypoxia, unrestricted growth of granulation tissue and endothelial cell-mediated absorption of bone matrix [13,14]. Gorham and Stout [7] hypothesised that trauma may trigger this disease process by stimulating the production of vascular granulation tissue. As in our case, the patient gave no such history of trauma except undergoing extraction of his teeth few months back.

Gorham's disease is usually monocentric and secondarily involves adjacent bones and soft tissue. However, multicentric maxillofacial cases have been reported by Tsang et al, [21] Oujilal et al [16] and Benhalima H et al [18] simultaneously affecting temporal bone, frontal bone, zygoma, mandible and maxilla. In the present case diffuse osteolysis of the right half of maxilla with cortical erosion was seen. The disease has been described as having a two-stage based on clinical presentation. Klein described 2 phases of disease and the first phase involves pain and swelling with associated massive osteolysis while the second one is the quiescent phase [22]. The duration of each phase may last for several weeks. The present case was in its first phase at the time of reporting and now has entered in to the quiescent phase.

In the present case the main complaint as the disease progressed was of severe pain with a burning quality which aggravated on palpation of the right maxillary region. This could be due to neuropathic pain involving the right maxilla or infraorbital nerve after osteolysis. This pattern of pain manifesting in this patient has not been reported in the previous case reports. The remaining teeth present were mobile with fistula and pus discharge.

The radiological findings are dramatic. A large localized region of bone destruction with possible involvement of contiguous bones, even with extension across joint spaces may be seen. Osseous structures are replaced with fibrous tissue. Associated soft tissue masses are rarely seen [17,19]. Moller [23] described 4-stage phases: the first stage or the early intraosseous stage with multiple ''patchy'' intramedullary and subcortical lucencies resembling osteoporosis. The second stage involves the coalescence of these radiolucencies. The third extraosseous stage, is characterized by cortical erosion and adjacent soft-tissue involvement. The fourth and final stage, the remaining bone is reabsorbed and finally replaced by fibrous tissue. The present case showed diffuse osteolysis of right maxilla extending to the nasal septum and palate.

Histopathological findings depend on the phase of the disease at the time of diagnosis. Two phases of the histopathological findings have been described in the literature by Heffez *et al*. [24] The first phase is increased vascular concentration in the bone-displacing fibrous tissue part and in the second phase only fibrous tissue is found. There were no angiomatous or fibrous areas seen in the present case but profuse bleeding was observed on surgical exploration.

Hefez listed criterias for diagnosing Gorham's disease and many authors used these criterias as a guide to diagnose the condition [24]. The criterias include positive biopsy showing angiomatosis, absence of tumour/cellular atypia, little or no osteoblastic response or dystrophic calcification, local progressive resorption, non-expansile, non-ulcerative lesion, absence of visceral involvement, osteolytic radiographic pattern and no hereditary, metabolic, immunologic or infectious cause. The present case fulfils most of the criteria as no cellular atypia, calcifications, expansion was seen. An osteolytic radiographic pattern was observed without any hereditary or metabolic disorder.

Gorham's disease should be included among the pathologic entities mimicking periodontal disease on radiograph, such as inflammatory disease like osteomyelitis, endocrine disorders such as hyperparathyroidism, intraosseous malignancies or metastases, lymphoma, infective process (e.g., tuberculosis and actinomycosis) and odontogenic tumours. Malignant disease must be ruled out histologically and lack of soft tissue mass rules out presence of neoplasm. Sclerosis and periosteal rection as seen in osteomyelitis are not observed in GD [25]. Other rare conditions such as histiocytosis X, mainly eosinophilic granuloma, may present with effects on other organ systems which differentiates it from Gorham's disease. Rosai-Dorfman disease (RDD) or sinus histiocytosis can also be included in the differential diagnosis which also presents with diffuse osteolysis and is accompanied by massive lymphadenopathy and constitutional symptoms [26].

The diagnosis is usually established by exclusion, where patients show unremarkable biochemistry and haematology findings, radiographic evidence of osteolytic lesions in association with histological proliferation of capillary and small lymphatic vessels [15]. In the present case the infectious etiology was ruled out by pus culture and no fungal organisms were detected. All the laboratory biomarkers were within normal limits thus ruling out metabolic and endocrine disorders. Absence of any ulcerative or expansile mass ruled out malignancy. Therefore, the diagnosis of Gorham's disease in the present case was by exclusion.

No consensus data has been found regarding definitive treatment. Some authors suggested the use of radiation therapy and the area irradiated should include the surrounding soft tissues that show clinical or imaging evidence of disease to arrest osteolysis and reduce capillary proliferation and occasional recalcification [27]. A case of radiation induced sarcoma has been reported in a 53-year-old male patient by Rodriguez-Vazquez JR et al after treatment of GD of the maxilla. Use of various analgesics, chemotherapy, biphosphonate [28], calcium, fluoride, alpha-2b interferon [29], vitamin D, hormones, amino acids, adrenal extracts, UV radiation, somatotrophin, and transfusions of placental blood or blood from growing young children have been reported resulting in limited success though cases with spontaneous recovery have also been reported. [30] In our case, the maxillary bone was stabilized after the surgical intervention and was advised Vitamin D and calcium supplements.

Conclusion

Gorham's disease is a rare type of idiopathic osteolysis with unknown etiology and specific pathogenesis. Diagnosis is by exclusion after ruling out infections, metabolic and malignant disease. Diagnostic X-rays and three-dimensional imaging are imperative for the diagnosis and follow up of Gorham's disease. The clinical appearance is often misleading, masquerading as infections and malignancies. Therefore, Gorham's disease should be included in the differential diagnosis of diffuse osteolytic lesions of the jaw.

AUTHOR CONTRIBUTION

S. No.	Work	Contributors	
		1	2
1.	Concept and design	Dr Deepa	
2.	Definition of intellectual content	Dr Deepa	
3.	Literature research	Dr Deepa	Dr Aravinda
4.	Clinical studies	Dr Deepa	
5.	Manuscript preparation	Dr Deepa	Dr Aravinda
6.	Manuscript editing and manuscript review	Dr Deepa	Dr Aravinda

Hosting Institution

This case study was carried out at Swami Devi Dyal Dental College and Hospital
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Conflict of Interest

The authors have no conflict of Interest to disclose

Declaration of interests

☒ The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

Patient Consent

Myself Dr Deepa Patil, Corresponding author of the manuscript has obtained consent from the patient for publishing the images. The patient's identity has not been revealed in the figires. The figures are comprised of Intra oral pictures and CT images.

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