

Vanishing Mandible: Report of a Rare Entity

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ABSTRACT

Vanishing mandible is an important manifestation of Gorham-Stout disease. GSD is a rare osteolytic bone disease of unknown etiology. It is characterised by massive osteolysis and gradual disappearance of bone. It may affect any bone of the body. Herein we report a case of vanished mandible in 16 years male with an emphasis on its differential diagnosis and a brief review of literature.

Keywords: Differential diagnosis; Gorham-Stout disease; osteolysis; vanishing mandible.

INTRODUCTION

Gorham's disease is a rare skeletal disorder which is characterised by massive osteolysis where the bone is destroyed and does not regenerate or repair and is replaced by fibrous connective tissue.¹ It is also known as Phantom bone disease, Vanishing bone disease, Gorham-Stout syndrome, Massive osteolysis, Idiopathic massive osteolysis, Progressive osteolysis, Massive Gorham osteolysis, Morbus Gorham-Stout disease (GSD). The disease is extremely rare with less than 200 cases reported. Jackson in 1838 was the first to describe a different entity of multiple osteolysis in humerus in 18 year old which was reported by Gorham and later by Gorham and Stout. Despite several studies its pathogenesis still needs to be clarified.² Herein we report a case of vanishing mandible in 16-year-old male.

CASE REPORT

A 16-year-old male reported with a swelling in lower jaw for one year which was not associated with pain. Swelling gradually increased and patient started to notice loosening of teeth in his lower jaw.

He also complaints of discharge of pus from his ear for one month. On extraoral examination there was facial asymmetry of lower half of face (Figure 1A). The swelling was firm on palpation extending two inches below the chin. Intraoral examination revealed mobile lower teeth, erythematous and edematous overlying mucosa with pain on palpation.

Cone beam computed tomography (CBCT) was done as a radiological assessment which revealed osteolytic lesion involving the mandible with osteolysis of the left body, ramus and coronoid and condylar process. Floating right mandibular teeth could also be noted. CBCT also revealed thinning of right lower border of mandible with osteolysis involving right ramus with thinning of coronoid and condylar processes (Figure 1B, 1C, 1D)

Based on clinical and radiographical findings provisional diagnosis of Vanishing bone disease was given. Mandibulectomy with reconstruction surgery was done (Figure 1A).

The resected mandible was received with condyle and entire dentition measuring 9X12cm along with right and left 1B lymphnodes (Figure: 2B).

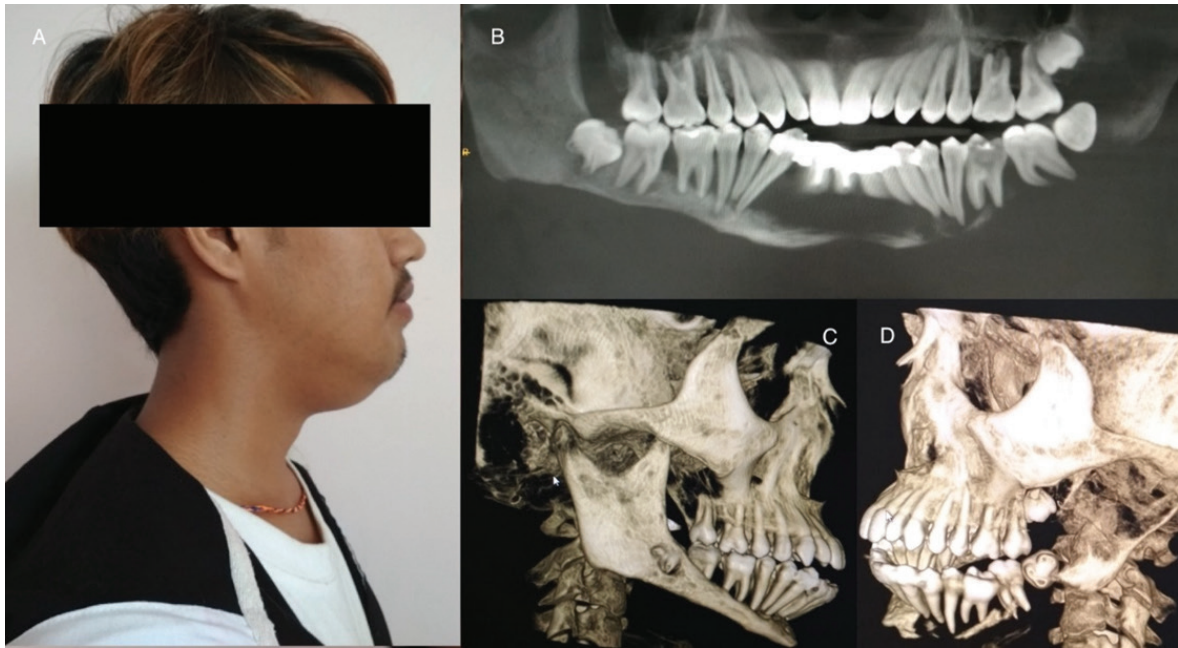


Figure 1A: Extraoral swelling involving mandible extending below the chin.
Figure 1B, 1C and 1D: Radiographic appearance of osteolytic lesion in mandible with floating teeth on the left side.

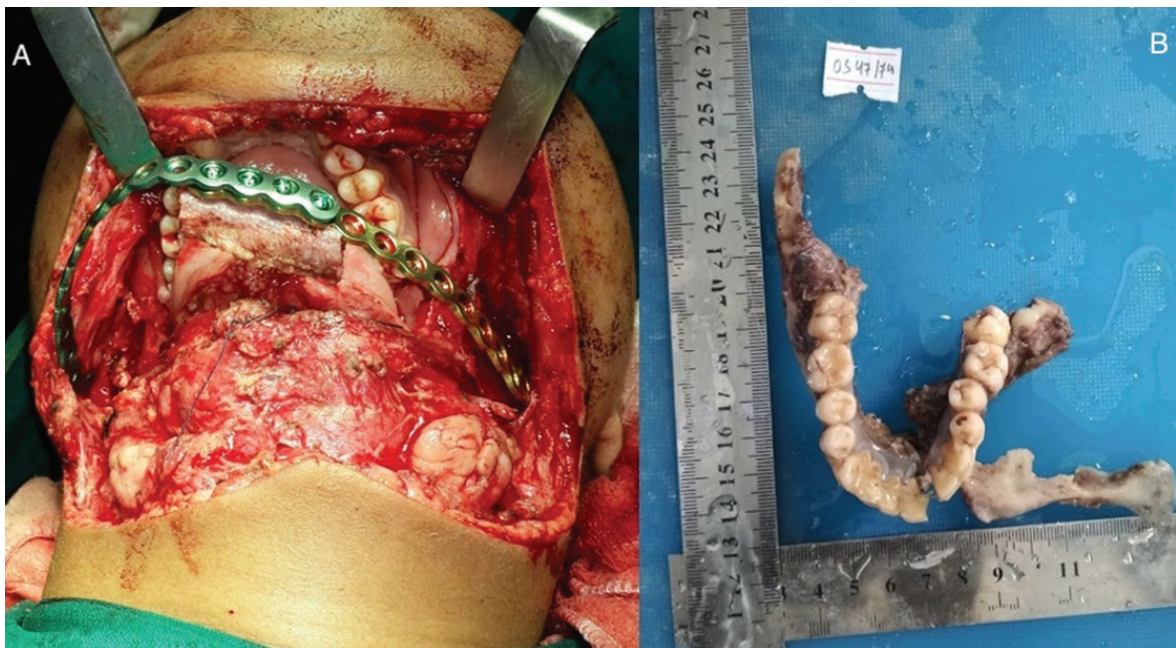


Figure 2A: Intra-operative picture of mandibulectomy followed by reconstruction surgery.
Figure 2B: Resected mandible with resorption of its maximum region.

Histopathological examination revealed fibrous connective tissue stroma showing numerous thin walled endothelial lined vascular spaces along with acute and chronic inflammatory cells infiltration with the extravasated red blood cells (RBCs) (Figure 3A). Right and Left 1B lymphnodes showed well defined capsule having normal architecture. Few areas showing acute inflammatory infiltration with inflammatory exudates and engorged blood vessels having extravasated RBCs (Figure 3B). Decalcified

section revealed lamellated bone with osteocytes within the lacunae. The surrounding stroma was loosely arranged with endothelial lined blood vessels (Figure 3C).

Osteomyelitis, giant cell lesions and eosinophilic granuloma were considered as differential diagnosis.² Osteomyelitis shows medullary spaces filled with inflammatory exudates with inflammatory cells chiefly polymorphonuclear

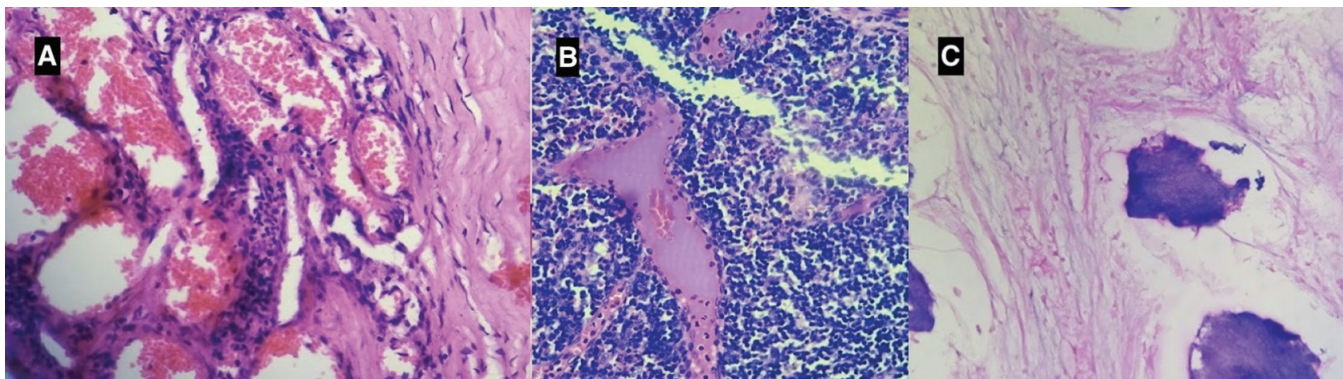


Figure 3A: Fibrous connective tissue stroma with numerous vascular component.

Figure 3B: Lymphonodes with acute inflammatory cell infiltration and inflammatory exudates.

Figure C: Decalcified section with lamellated bone with osteocytes within the lacunae with loosely arranged stroma filled with endothelial lined blood vessels.

leukocytes, but may show occasional lymphocytes and plasma cells. Osteoblast cells rimming the bony trabeculae are generally destroyed and trabeculae may lose their viability and begin to undergo slow resorption none of the features were noted in our case hence ruling out osteomyelitis.³

Similarly, giant cell lesions included were central giant cell granuloma, giant cell tumor, cherubism, brown tumor of hyperparathyroidism, aneurysmal bone cyst. Central giant cell granuloma shows numerous multinucleated giant cells embedded in a fibro-cellular stroma having spindle-shaped fibroblasts.⁴ Foci of hemorrhage with hemosiderin pigment along with newly formed osteoid could be observed in the stroma which was not present in our case. Whereas giant cell tumor is composed of cellular stroma comprising of plenty of evenly distributed multinucleated giant cells with agglomeration of around 25-50 nuclei in center surrounded by clear cytoplasm. The proliferating stromal cells are hyperchromatic with pleomorphic areas of pathologic bone resorption.⁵

Histologically cherubism shows features of central giant cell granuloma with fewer giant cells and perivascular cuffing which were not the features seen in our case.⁶ Brown tumor of hyperparathyroidism is composed of multinucleated giant cells with areas of hemorrhage, deposition of hemosiderin pigment, irregular trabeculae of woven bones lined by osteoblast and stroma with dense collagen fiber and spindle cells.⁷ Lastly aneurysmal bone cyst shows cystic space without epithelial lining loosely arranged collagen fibers with fibroblasts,

cavernous or sinusoidal blood filled spaces, areas of hemorrhage with multiple giant cells and chronic inflammatory cell infiltration.⁸ Since all these features were not seen in our case giant cell lesions were ruled out of the differential diagnosis.

Eosinophilic granuloma in the histopathology shows infiltration by Langerhans, eosinophils, giant cells, neutrophils, foam cells, lymphocytes, plasma cells, fibrosis, necrosis and may have typical and atypical mitotic figures.⁹ None of the above described histological features were present in our case.

Thus, based on clinical, radiographical and histopathological features final diagnosis of Vanishing bone disease (Gorham-Stout disease) was given.

DISCUSSION

Gorham disease is rare phenomenon which is described as gradual destruction of single or more bones, replaced with expanding vascular component subsequently replaced by fibrous connective tissue without bone regeneration. Commonly involved sites include mandible, ribs, scapula, humerus, pelvis, and femur. There is no sex or racial predilection, and this disease is and mostly seen below the age of 40 years.²

According to Klein the disease has two phase, first phase involves pain and swelling with osteolysis and second one is the quiescent phase.² Similarly, Moller described the syndrome with four phase. First stage or the early intraosseous stage showing

by multiple patchy intramedullary and subcortical radiolucencies resembling osteoporosis. Second stage shows the coalescence of these radiolucencies with development of new radiolucent areas at the periphery of the involved region. Third stage or the extraosseous stage, is characterized by cortical erosion and adjacent soft tissue involvement. Fourth and final stage is characterized by resorption of the remaining bone with replacement by fibrous tissue. Resorption of the bone results in tapering of the involved end giving the appearance as “sucked candy.” According to Moller staging of GSD, present case was in fourth and final stage.²

Etiology of this disease remain unclear. However, there are many hypotheses of the etiologies including post-traumatic hyperemia, changes in blood pH, hypoxia, unrestricted growth of granulation tissue and endothelial cell-mediated absorption of bone matrix.¹⁰ Gorham and Stout hypothesized that trauma may initiate this disease process by stimulating the production of vascular granulation tissue, while Devlin suggested the role of Interleukin -6 (IL-6) in the development of this disease. As IL-6 is produced by osteoclast or in response to muscular cells inducing bone resorption through osteoclastic activity. Cells of monocyte – macrophage lineage may initiate the pathogenesis accelerating angiogenesis and osteoclastogenesis.² Heyden et al. suggested that local hypoxia and lowering of pH is produced due to the slow circulation, which favors the activity of various hydrolytic enzymes. They observed strong activity of both acid phosphatase and leucine aminopeptidase in mononuclear perivascular cells that were in contact with remaining bone, perhaps indicating that these cells are more important in process of osseous resorption.²

Histopathologically these lesions are characterized by the presence of proliferating thin walled vascular spaces of varying size, later which is replaced

by fibrous connective tissue.¹ Hefez et al. has postulated criteria for diagnosis of vanishing bone disease which include histopathology showing angiomatosis without cellular atypia, few or totally absence of osteoblastic response, non expansile and non-ulcerative lesion with local progressive resorption and no dystrophic calcification with radiograph showing osteolytic pattern. There should be no familial history and absence of metabolic and infectious cause.² Since all these features were appreciated in our case the diagnosis was given as GSD.

Lesion stains for CD31, CD34 and Factor VIII confirming a vascular origin of while Ki67 index show low cellular proliferative activity which implies a nonmalignant proliferation. Immunohistochemical investigations including CD20 and CD79a which confirmed B lymphocytes, CD3 showed T-lymphocytes, kappa and lambda light chains confirmed polyclonality of plasma cells and PGM1 showed presence of neutrophils and foamy histiocytes. In view of the presence of many inflammatory cells, absence of malignant cells and evidence of blood and lymphatic vessel proliferation, the hypothesis of Gorham’s disease is considered.²

Treatment include surgery using bone grafting. Radiation therapy is reported to be effective in arresting osteolysis. Radiation therapy, anti-osteoclastic medication (bisphosphonates), alpha-2b interferon, adrenal extracts, androgen are also the treatment of choice.²

Gorham Stout disease is a rare musculoskeletal disease whose etiology is still uncertain with long list of differential diagnoses. It is therefore important for clinicians and pathologist to be aware of its existence as a rare case in maxillofacial skeleton whose diagnosis is to be made based on exclusion of other diseases.

REFERENCES

1. Ravindran R, Karunakaran A. Case Report Gorham ' s Disease of the Mandible. *Case Rep Dent.* 2013;2013:3–6.
2. Ibrahim N, Kipli NP, Toha A, Ismail R. Gorham ' s Disease of a Mandible : A Case Report. *Cosmetol Oro Facial Surg.* 2015;1(1):103-6.
3. Rajendran A, Sivapathasundharam B. *Shafer's Textbook of Oral Pathology.* 6th ed. Delhi; Elsevier India: 2009. 493-98 p.
4. Alchalabi N, Salih H, Merza A. Central giant cell granuloma of the maxilla: a case report. *Int J Dent Res.* 2017;5(2):145-7.
5. Giri GVV, Sukumaran G, Ravindran C, Narasimman M. Giant cell tumor of the mandible. *J Oral Maxillofac Pathol.* 2015;19(1):108.
6. Jiao Y, Zhou M, Yang Y, Zhou J, Duan X. Cherubism misdiagnosed as giant cell tumor: a case report and review of literature. *Int J Clin Exp Med.* 2015;8(3):4656-63.
7. Shetty AD, Namitha J, James L. Brown tumor of mandible in association with primary hyperparathyroidism: a case report. *J Int Oral Health.* 2015;7(2):50-2.
8. Marin Fernandez AB, Garcia Medina B, Martínez Plaza A, Aguilar-Salvatierra A, Gomez-Moreno G. Aneurysmal bone cyst of the mandible affecting the articular condyle: a case report. *Clin Case Rep.* 2016;4(12):1175–80.
9. Mani V, Vinod VS, George A, Sivaprasad KK. Eosinophilic granuloma of mandibular condyle: resection and complete regeneration. *J Maxillofac Oral Surg.* 2015;14(1):107–10
10. Asha ML, Jose AA, Babshet M, Bajoria AA, Naveen S. Vanishing bone disease of the mandible - A case report. *Gerodontology.* 2013;30(4):296–302.