

Vanishing bone disease: An enigma

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Abstract

Vanishing bone disease is a rare clinical entity with unknown etiology. This disease affects individual irrespective of age or sex. Various names have been used in the literature to describe this condition such as Gorham's disease, phantom bone disease, massive osteolysis, disappearing bone disease and acute spontaneous absorption of bone. The pathogenesis is unknown and the treatment still remains controversial. Considering the rarity of the disease, we report here an interesting and unique case of massive osteolysis of the lower jaw that affected the mandibular basal and alveolar bone. The diagnoses lead on the association of clinical, radiological and histological features.

Keywords: Gorham's disease, mandible, massive osteolysis, vanishing bone disease

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INTRODUCTION

Vanishing bone disease (VBD) is an uncommon disorder characterized by the proliferation of vascular channels that result in the destruction and resorption of osseous matrix.^[1] It was first described in 1838 by Jackson, who reported a case of "boneless arm." The first case reported in jaws was by Romer in 1924 in a 31-year-old woman. It was Gorham in 1954 and Gorham and Stout in 1955, they presented a case series and defined the condition as a pathologic process.^[2] No evidence of a malignant, neuropathic or infectious component was found in the causation of this disorder.^[1]

Although massive osteolysis has been documented in patients up to 70 years of age, the most affected patients are children and young adults. About 50% of all patients report an episode of trauma before the diagnosis that is often trivial in nature.^[3] Any bone can be affected, although

there is a predilection for the pelvis, humerus, axial skeleton and mandible.^[2]

The case presented here was characterized by massive idiopathic osteolysis exclusively affecting the mandible. The clinical, radiological and histological findings were consistent with the criteria for VBD.

CASE REPORT

A young female patient presented with a chief complaint of pain and facial deformity of the left side of the face over a period of 6 months. Past medical, dental and family history of the patient was noncontributory. There was no history of trauma, sinus opening or pus discharge. Pain was moderate, continuous in nature and radiating to the left side of the face and neck. Extraoral examination revealed facial asymmetry due to the step deformity in the left body of the mandible in the premolar molar region. Depression

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was felt in the left preauricular region with deviation of the mandible on the left side during the opening of the mouth. The left inferior border of mandible and ramus was not palpable [Figure 1]. Intraoral examination showed step defect in 35 and 36 regions with segmental mobility with 36, 37 and 38. Ascending ramus of the left mandible was not palpable [Figure 2].

Investigations

Radiographic examination revealed radiolucency in 36, 37 and 38 regions with their displacement and root resorption. The left ramus and the inferior border of the mandible were not traceable from 35 regions. No bony support was seen with 36, 37 and 38 right side of the mandible up to 34 appeared normal [Figure 3]. Computed tomography (CT) scan of mandible showed lytic expansile destructive lesion of the left hemimandible with cortical destruction and associated soft-tissue swelling with loosened teeth lying in the matrix of the lesion with submental lymphadenopathy. Only a small part of the

condyloid process was spared which was displaced into the anterior infratemporal fossa [Figures 4 and 5]. Based on the CT findings, the possibility of eosinophilic granuloma, VBD, round cell tumor was raised. Hematological and biochemical assessment (Acid and Alkaline Phosphatase) were all normal. The findings of pelvic and chest radiograph were normal.

Microscopy

Mandibular bone biopsy adjacent to the zone of osteolysis on the left side was performed. Hematoxylin and eosin stained section showed lesional tissue composed of connective tissue stroma containing sparsely arranged collagen fibers with few dense collagen fiber bundles. Numerous small and big blood vessels were seen with few showing thickened vessel wall. A dense chronic inflammatory cell infiltrate composed of lymphocytes and

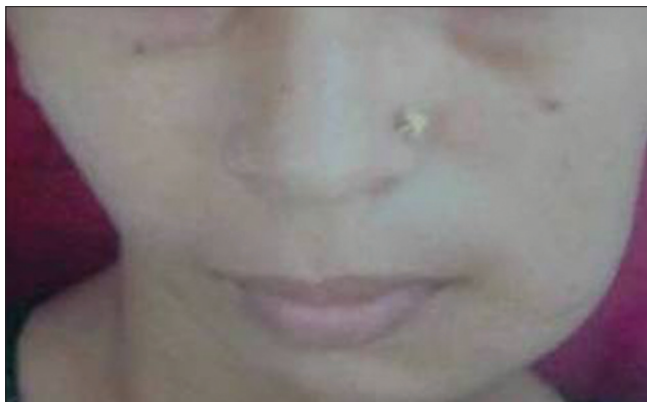


Figure 1: Extraoral examination revealed facial asymmetry due to the step deformity in the left body of the mandible in the premolar molar region. The left inferior border of mandible and ramus were not palpable



Figure 2: Intraoral examination showed step defect in 35 and 36 regions with segmental mobility with 36, 37 and 38. Ascending ramus of the left mandible was not palpable



Figure 3: Radiolucency in 36, 37 and 38 regions with their displacement and root resorption. Left ramus and the inferior border of the mandible are not traceable from 35 regions. No bony support is seen with 36, 37 and 38. The right side of the mandible up to 34 appears normal

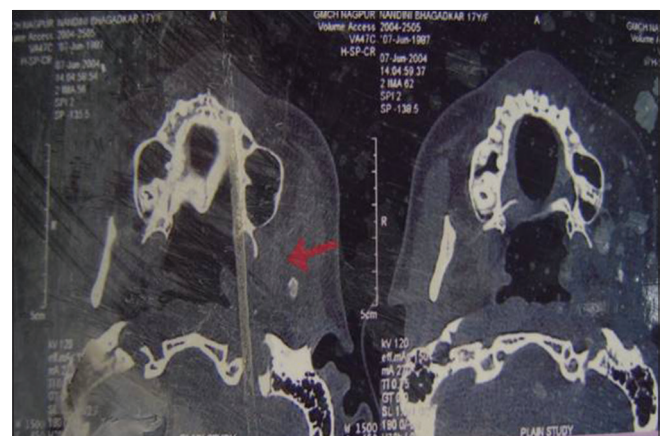


Figure 4: Computed tomography scan of mandible showed lytic expansile destructive lesion of the left hemimandible with cortical destruction. Only a small part of the condyloid process is spared which is displaced into the anterior infratemporal fossa

abundant plasma cells was evident. Lesional tissue was covered by stratified squamous epithelium [Figures 6 and 7].

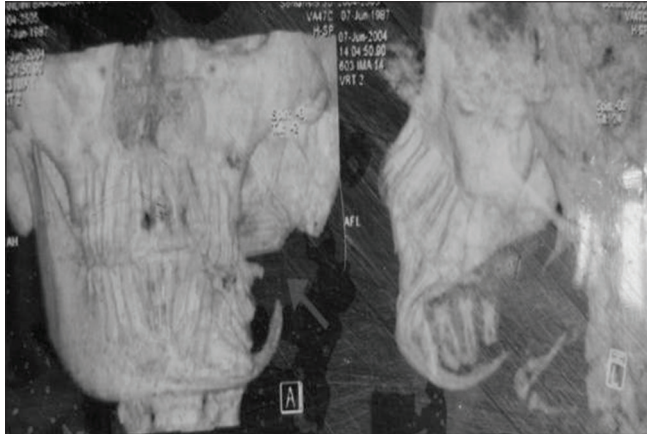


Figure 5: Complete absence of the left mandible

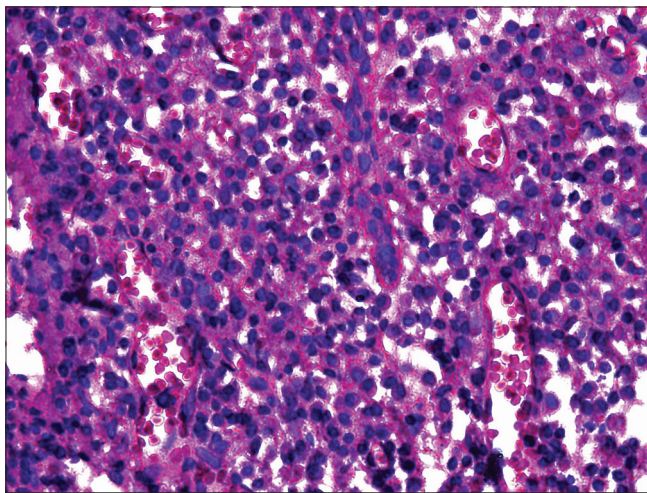


Figure 6: Numerous small and big blood vessels were seen with few showing thickened vessel wall. A dense chronic inflammatory cell infiltrate composed of lymphocytes and abundant plasma cells was evident

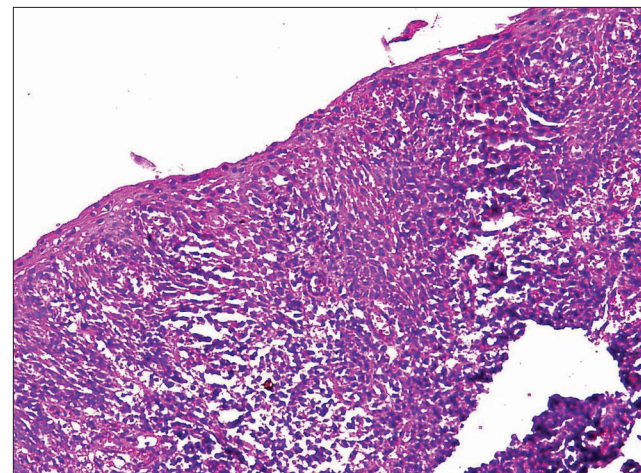


Figure 7: Lesional tissue was covered by stratified squamous epithelium

DISCUSSION

VBD, as the name suggests, is a condition in which any bone starts vanishing or disappearing. The word “vanishing” has been used with respect to the disease’s radiographic presentation.^[4] It is characterized by uncontrolled, destructive proliferation of vascular or lymphatic capillaries within bone and surrounding soft tissue.^[5] It affects patients ranging widely in age, from 2 months to 78 years, but most common in those under 40 years. Different theories have been suggested to explain the pathogenesis of the disease, which include abnormal proliferation of vascular tissue; activation of silent hamartoma by minor trauma; hydrolysis by enzymes due to local hypoxia and acidosis; It is found that signaling pathway of the Platelet-Derived Growth Factor receptor BB and cell of monocyte-macrophage lineage which fastens osteoclastogenesis and angiogenesis plays a key role in the pathogenesis of massive osteolysis.^[6] The nonspecific complaints, lack of markers of systemic illness and rarity of the disease contribute to delayed diagnosis.^[7]

Heffeze *et al.* described the criteria for the diagnosis of massive osteolysis as:^[2]

- Evidence of local progressive osseous resorption
- Minimal or no osteoblastic response and an absence of dystrophic calcification
- Nonexpansile, nonulcerative lesion
- Absence of visceral involvement
- Osteolytic radiographic pattern
- Negative findings for a hereditary, neoplastic, immunologic, infectious and endocrinologic origin.

Our case features most of the criteria put forth by Heffeze *et al.*

In approximately, 30% of affected patients, maxillofacial involvement is noted.^[8] The mandible being the most frequently affected with sixty cases reported in the literature. The main symptom is intermittent pain and swelling.^[9] Other symptoms include mobile teeth, malocclusion, deviation of the mandible and clinically obvious facial deformity. Pathologic fracture of the mandible may also occur.^[3] All these symptoms were evident in our case also.

In general, the result of laboratory investigation in these patients is completely within normal limits,^[3] which was documented in our case as well. The elevated interleukin-6 levels reflected the osteoclastic activity due to their sensitivity to certain growth factors.^[8]

The earliest radiographic sign of the disease has been reported to be one or more intermedullary subcortical radiolucencies of variable size, usually with distinct margins and thin radiopaque borders. A characteristic tapering occurs when long bones are affected.^[10] Literature shows radiographs, CT, and magnetic resonance imaging all can be used in the diagnosis of this entity. Panoramic radiograph shows resorption and decreased vertical height of the mandibular body with the resorption extended toward the basal bone.^[8]

The characteristic histopathology is the replacement of bony tissue by intrafibrous connective tissue containing many thin-walled vascular channels, including lymphatic capillaries termed histologically as hemangiomas.^[11] Osteoclastic activity in adjacent bone fragments is usually not conspicuous.^[3]

Radiation therapy, anti-osteoclastic medications (bisphosphonates) and alpha-2b interferon constitute the treatment for this disease. Surgical treatment includes resection of the lesion and reconstruction using bone grafts and prostheses.^[8] In most cases, bone grafts tend to undergo resorption and are not helpful. Unfortunately, no single treatment modality has proven effective in arresting the disease.^[1]

The fate of these patients is difficult to predict. In most cases, bone resorption ceases spontaneously after some years, regardless of treatment.^[9] A high morbidity and mortality is seen in patients with spinal or visceral involvement.^[1]

The clinical significance of VBD is, it should be included among the pathologic entities mimicking periodontal disease on radiograph, such as inflammatory disease (e.g., osteomyelitis), endocrine disease (e.g., hyperparathyroidism), intra-osseous malignancies or metastases, lymphoma, histiocytosis X, mainly eosinophilic granuloma, infective process (e.g., tuberculosis and actinomycosis) and odontogenic tumors.^[12] Progressive and unusual substantial bone destruction without evidence of repair is almost pathognomonic for VBD.

To conclude, when VBD involves the mandible, the role of the periodontologist is extremely important in diagnosing

and preventing the functional and aesthetic consequences of extensive bone loss. Although clinical and radiological findings are key to diagnosis, a histopathological report is mandatory, to exclude other disease processes.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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