CASE REPORT

Gorham's disease: a report of a case with mandibular involvement in a 10-year follow-up study

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Gorham's disease, or vanishing bone disease, is a rare condition of unknown aetiology. Any bone of the body can be affected, although there is a predilection for the pelvis, humerus, axial skeleton and the mandible. The mandible is the most commonly involved bone in the maxillofacial region. 41 cases have been reported so far in the literature showing involvement of the mandible in Gorham's disease. This paper presents a rare case of Gorham's disease involving the mandible in a 38-year-old male with a 10-year follow-up.

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Introduction

Gorham's disease, also known as massive osteolysis, is a rare disorder characterized by spontaneous and progressive osteolysis of one or more skeletal bones.¹ Jackson² was the first person to describe a case of vanishing bone disease in 1838 while Romer³ was the first to report a case in the jaws of a 31-year-old female in 1924. It was Gorham⁴ in 1954 and Gorham and Stout¹ in 1955 who presented a case series and defined the condition as a specific pathological process. To date, approximately 200 cases have been described in the literature, with 41 cases involving the maxillofacial region, especially the mandible.⁵

The aetiology of this disease remains unknown, although an initial trauma or modification of local conditions, such as variation in pH, inflammations etc., are suspected. It is common in children and young adults; however, cases are reported in patients aged up to 70 years or older. There is no racial predilection, although there is a gender predilection in males. The disease can be monostotic or polyostotic, although multicentric involvement is exceptional.⁶ Pain, malocclusion and deformity have been reported in 30% of the cases with maxillofacial involvement.⁷

Gorham's disease may be associated with major dental and maxillofacial complications and radiographic

assessment plays an important role in the identification of massive osteolysis despite the clinical manifestations. Herein, we report a case of monocentric massive osteolysis involving the mandible with its distinctive radiographic manifestation in a 10-year follow-up study.

Case report

A 38-year-old male patient reported to the dental department at the MS Ramaiah Dental College and Hospital, Bangalore, Karnataka, India, in February 2001 with a complaint of mobile teeth in the left mandible. The patient was asymptomatic with no history of trauma, swelling, sinus opening or pus discharge. The medical history was insignificant and the patient was otherwise in good general health. The family and personal histories were non-contributory. On intraoral examination, periodontal pockets in relation to right and left lower posterior teeth and Grade III mobility with respect to the lower left first, second and third molar was observed. A panoramic radiograph revealed generalized horizontal bone loss (Figure 1) and a diagnosis of chronic generalized periodontitis was made. The mobile teeth were extracted in the Department of Oral and Maxillofacial Surgery, MS Ramaiah Dental College and Hospital, and the sockets healed uneventfully. Oral prophylaxis and curettage was performed.

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Figure 1 Panoramic radiograph showing alveolar bone loss in the lower left quadrant

3 years later in 2004, the patient reported with a complaint of deviation of the mandible to the left side on mouth opening. Clinical examination revealed atrophy of the alveolar ridge in relation to the lower left quadrant. The occlusion was normal (Angle's Class I occlusion) and on opening there was deviation of the mandible to the left side. There was no history of weight loss, paraesthesia, motor nerve deficits or palpable lymph nodes. A panoramic radiograph revealed resorption and decreased vertical height of the left mandibular body and resorption of the left condyle. The resorption extended medially from the distal surface of the left second premolar, distally from the anterior border of the ramus of the left mandible and inferiorly about 1.5 cm from the inferior border of the mandible. The angle of mandible was intact (Figure 2). A three-dimensional CT scan substantiated the above-mentioned findings (Figure 3). The findings of the pelvic and chest radiography were normal. No bony sequestra or subperiosteal bone deposition indicative of osteomyelitis were present. Blood chemistry values did not reveal any abnormality. The overall

Figure 2 Panoramic radiograph taken 4 years later shows atrophy of the ridge with decreased vertical height. Osteolysis is seen extending to the left condyle and body of the mandible

clinical presentation, radiological features and biochemical findings favoured the diagnosis of Gorham's disease of the left mandible. Incisional biopsy was advised but the patient was not ready to undergo the procedure. The treatment plan included surgical intervention with excision and subsequent bone grafting, but the patient was unwilling to undergo any treatment procedure. The patient was advised to take vitamin D and calcium supplements for a period of 6 months. Regular follow-up was recommended.

Follow-up imaging from 2004 to 2008 confirmed further resorption of the mandible involving the condyle, ramus, angle of the mandible, body and basal bone of the left side of the mandible over a period of 4 years. Progressive resorption has led to tapering of the remnant mandibular bone (Figure 4). A three-dimensional CT scan was performed which showed marked alveolar bone loss of the left mandible involving the condyle, ramus, angle and decrease in height of the body of the mandible (Figure 5). Radiographic examination during March 2010 showed no change or progression from the existing lesion which was observed in 2008 (Figure 6).

a

b

Figure 3 (a and b) Three-dimensional CT scan revealing atrophy of the alveolar ridge of the left body mandible and condyle. Angle of mandible is intact

Figure 4 Panoramic radiograph shows osteolysis of the remnant basal bone

Spontaneous arrest of the osteolytic process involving the left mandible occurred.

Discussion

Among the many cases of craniofacial tissue involvement described in literature, the mandible is the most frequently involved site.⁵ According to Schiel and Prein,⁷ only 31 cases have maxillofacial involvement meeting the Heffez criteria.¹² These criteria include the evidence of local progressive osseous resorption, minimal or no osteoblastic response, absence of dystrophic calcification, non-expansile, non-ulcerative lesion, absence of visceral involvement, osteolytic radiographic pattern, positive biopsy for angiomatosis and negative findings for a hereditary, metabolic, neoplastic, immunological or infectious origin.⁸

Gorham's disease is usually monocentric and starts in a particular bone but has been reported to secondarily involve adjacent bones and soft tissue. It is characterized by progressive loss of bony structures with replacement by thin-walled capillary-sized channels and later by fibrous connective tissue. Gorham and Stout¹ observed replacement of bone by connective

tissue containing many dilated capillaries or anastomosing vascular spaces filled with blood cells. Progression of the disease results in dissolution of cortical and cancellous bone until fibrotic bands replace all bony tissue. Proliferation of vascular elements was reported to be a prominent feature associated with massive osteolysis. According to Gorham and Stout,¹ massive osteolysis is not the result of osteoclastic activity but is related to active hyperaemia, mechanical forces, changes in pH and other unknown causes.

Gorham's disease is characterized by post-traumatic or spontaneous bone destruction that appears to be related to an active hyperaemia of bone. The diagnosis of Gorham's disease is difficult, particularly in the early stage. In the early stages, it must be differentiated from osteolysis associated with infection, such as osteomyelitis or infectious rheumatoid arthritis, trauma, endocrine diseases such as hyperparathyroidism, intra-osseous malignancies, metastasis, eosinophilic granuloma and osteolysis associated with diseases of the central nervous system, such as tabes dorsalis, syringomyelia, leprosy or myelodysplasia.⁹

Most cases occur in children and adults under the age of 40 years, although it has been reported in patients aged from 18 months to 72 years. Our patient reported at the age of 38 years with a complaint of mobile teeth in the left mandibular quadrant. The onset is often insidious and pain is not usually a feature unless pathological fracture develops.⁶ Mobile but vital teeth with accompanying gingival haemorrhage, hypoplasia, pain, malocclusion and resorption of the affected alveolar and adjacent bone may be the manifestations of maxillofacial involvement of Gorham's disease. Involvement of the temporomandibular joint by Gorham's disease can be mistaken for temporomandibular dysfunction.⁷

The most striking aspect of Gorham's massive osteolysis is its radiographic appearance. Radiographic findings of patients with Gorham's disease were first described by Johnson and McClure in 1958.¹⁰ During the initial stages; the radiolucent foci appear in the

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Figure 5 (a and b) Three-dimensional CT scan revealing marked alveolar bone loss of the left side of the mandible involving the condyle, ramus, angle and body of the mandible with tapering of the remaining basal bone

Figure 6 Panoramic radiograph showing spontaneous arrest of the osteolytic process

intramedullary or subcortical regions with undisturbed margins. Subsequently, these foci coalesce and eventually involve the cortex before slow progressive resorption, dissolution, fracture, fragmentation and disappearance of a portion of the bone. Progressive involvement reveals “tapering” or “pointing” of the remaining osseous tissue and atrophy of soft tissues. In our case, the earliest signs were tooth mobility with crestal bone loss, eventually progressing to involve the condyle, ramus, angle and body of the left mandible with tapering of the remnant mandibular basal bone.¹⁰ Frederiksen et al¹¹ observed that the mandible is more frequently involved than the maxilla with varying degrees of involvement of adjacent structures, including the hard palate, sphenoid bone, zygomatic bone etc. The progressive resorption of the mandible and the lytic process progress until the wispy remnants of the bone remain, at which point there is a radiographic absence of bone. CT plays a major role in evaluating the involvement of facial bones and the skull and can determine the extension into the base of the skull.¹² A radioisotopic scan may demonstrate increased vascularity in the initial images and subsequently an area of decreased uptake corresponding to the site of diminished or absent osseous tissues.¹³

In the review study by Anavi et al,¹⁴ the mean duration of symptoms such as local pain, mobile teeth, malocclusion and facial deformity before diagnosis was

6.4 years. In most cases, bone destruction progresses over months to a few years and results in the total loss of the affected bone or bones. Some patients, however, may experience a spontaneous arrest of the process without complete loss of the affected bone. In our case, more than two-thirds of the left mandible resorbed over a period of 6–7 years, following which spontaneous arrest of the osteolytic process was observed during the follow-up period (2008–2010).

The prognosis varies from slight disability to fatal outcome in some cases, approximately 16%.¹⁵ Severe disability results from the involvement of the cervical spine, pelvis and thorax. Long-term survival with varying deformation or disability is possible with peripheral bone disease.^{16,17}

Management of Gorham's disease is a challenge for clinicians. Different forms of therapy have been described, including medical treatment with oestrogen, calcium, vitamin D, fluoride, bisphosphonates and calcitonin,^{18,19} as well as surgical excision, bone grafting and prosthetic implants.^{20,21} Success of the above reported treatments have been very limited. Low-dose radiotherapy has recently been suggested to arrest the course of osteolysis in Gorham's disease; however, radiotherapy is still controversial and not widely accepted as the chances of radiation induced maldevelopment and malignant transformation are high, especially in children.^{22–24}

Conclusion

Massive osteolysis or Gorham's disease is a silent disease with potential complications when afflicting the maxillofacial region. As the clinical symptoms are not characteristic of the disease and may mask the underlying perilous disorder, panoramic radiographs and the various imaging modalities play an imperative role in the diagnosis of this condition. No treatment to date has proven to be effective in this disorder. Dental surgeons play a pivotal role in identifying this rare disorder and it should be included in the differential diagnosis of any obscure osteolytic process.

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