Rare disease

Progressive, massive osteolysis of mandible (Gorham's disease): report of a case

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Summary

A rare case of progressive osteolysis of mandible is discussed with clinicoradiological presentation. The purpose of the case report is to make the medical community aware of this rare and fascinating disorder in the maxillofacial region. The disease is characterised by spontaneous progressive osteolysis of one or more skeletal bones. Occurrence in maxillofacial region is rare with fewer than 35 reported cases affecting the facial and jaw bones. The present case report is about the mystery of bilaterally missing ascending rami. An attempt is made to draw attention of the medical fraternity to this unusual and rare entity which can induce instability, dysfunction and cosmetic disturbances.

BACKGROUND

Progressive osteolysis of mandible is a rare entity affecting the maxillofacial region with fewer than 35 cases reported in international literature till date. The disease is poorly understood and often gets misdiagnosed as very few clinicians get the opportunity to diagnose and treat this type of disorder. The purpose of the following case report is to make the medical community aware of this rare and fascinating disorder of the musculoskeletal system. We would also like to emphasise the need to investigate the head and neck region when examining a case of Gorham's disease since mandible is a high risk location of the disease due to probability of involvement of base of skull and spine.

CASE PRESENTATION

Complaint and history

A well-built 70-year-old man reported with a chief complaint of mobility of teeth and difficulty in chewing. The patient was unaware of any preexisting facial deformity. His expectation was total extractions and complete denture. Medical history and family history were not contributory. Moreover, there was no history of trauma.

Clinical assessment

Extraorally atrophic and retrognathic mandible was noticed. There was bilateral depression in the pterygomasseteric area. The distance between menton and hyoid bone was remarkably less. There was no neurological deficit of inferior

Figure 1 Right facial profile of patient.

Figure 2 Left facial profile of patient.

Figure 3 Pre-extraction orthopantomograph of patient.

alveolar nerve. On palpation, the inferior border of the mandible could not be traced posterior to the angle of mandible (figures 1 and 2). There was a small depression in the parasymphiseal area. No mass or lymph nodes were palpable. TM joint could not be palpated on either side. The mandible was hyper mobile and had a hanging jaw appearance. Entire mandible could be manipulated backwards and laterally freely. However there was restricted mouth opening.

Intraorally periodontally compromised dentition with multiple missing teeth was seen. There was anterior open bite with gagging of posterior teeth. Occlusion could not be established. There was no evidence of expansile or ulcerative lesion.

INVESTIGATIONS

Initial examination

Radiologic investigations

Orthopantomograph (OPG) showed that there was bilateral absence of ascending rami and coronoid processes on either side of the mandible. The right mandibular condyle was also not visible whereas the left one, though present, was rudimentary and rotated anteriorly and superiorly. There was pronounced resorption in the parasymphiseal area (figure 3). CT scan with 3D reconstruction confirmed the same findings (figures 4 and 5).

Radiology of long bones, chest and pelvis showed no involvement of other bones.

Figure 4 CT scan showing mandibular resorption on the right side.

Laboratory investigations

Haemogram, blood sugar level, serum calcium, serum alkaline phosphatase serum acid phosphatase, parathyroid hormone, erythrocyte sedimentation rate and rheumatoid arthritis factor were all within normal limits.

Histopathology

Microscopic study showed fibrocollagenous tissue with tiny bit of skeletal muscle tissue. Many congested blood vessels were seen with sparse mononuclear infiltrate all over. There was no evidence of tuberculosis or malignancy. However, few osteoclasts found.

DIFFERENTIAL DIAGNOSIS

The following differential diagnosis of osteolytic diseases was considered before arriving at a final diagnosis:

1. Massive osteolysis (Gorham's disease)
2. Idiopathic multicentric osteolysis
3. Multicentric osteolysis (Carpal and tarsal osteolysis)
4. Hereditary multicentric osteolysis
5. Neurogenic osteolysis
6. Osteolysis due to neoplastic, infectious and immunologic diseases.

TREATMENT

The patient was offered treatment consisting of radiation followed by bisphosphonate therapy and finally reconstructive surgery. The patient was completely asymptomatic and hence he refused any form of treatment. Therefore total extraction was carried out following which he was lost to follow-up (figure 6).

DISCUSSION

An entity with idiopathic osteolysis has a number of eponyms like vanishing bone disease, lymphangiomatosis, massive osteolysis, phantom bone disease and Gorham's disease.¹ The first case was reported by Jackson and described the spontaneous resorption of a humerus and led him to title his work 'A boneless arm'. The disease is characterised by spontaneous progressive osteolysis of one or more skeletal bones.² Occurrence in maxillofacial region is rare with fewer than 35 reported cases affecting the facial and jaw bones.³ The present

Figure 5 CT scan showing left side of resorbed mandible.

case report is about the mystery of bilaterally missing ascending rami. An attempt is made to draw attention of the medical fraternity to this unusual and rare entity which can induce instability, dysfunction and cosmetic disturbances.

Clinicians' awareness and a higher degree of clinical suspicion are required to arrive at a definitive diagnosis in osteolytic lesions. Many cases of massive osteolysis are described in literature with a variable presentation but very few are reported involving maxillofacial bones. The disease is notoriously difficult to diagnose and is often diagnosed through a process of elimination of other osteolytic disorders.

Figure 6 Orthopantomograph of patient after undergoing total extraction.

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- Neurogenic osteolysis
- Osteolysis due to neoplastic, infectious and immunologic diseases.

Radiographic examination plays an important role in the diagnosis of massive osteolysis regardless of clinical symptoms.⁵ In radiographic study of mandibular osteolysis Kriens defined the disappearance of cortical bone, porosis of trabecular structures, disappearance of lamina dura and atrophy of bone contour as characteristic findings of Gorham's disease involving the mandible.⁶ Disease process may extend to contiguous bones. The radiographic picture of the present case as displayed on OPG and 3D reconstructive CT scans showed bilaterally missing ascending rami with rudimentary condyle on the left side. Intraosseous resorption of alveolar bone and destruction of cortex around masseter and medial pterygoid muscle induced restriction of mouth opening.

A series of laboratory investigations were carried out but they failed to detect osteolysis secondary to:

1. Pathology (cyst, tumour, infection)
2. Metabolic and endocrinal disorders
3. Neoplastic disease
4. Trauma and
5. Connective tissue disorders.

Rapid review of literature revealed that in Gorham's disease, surprisingly all the laboratory investigations were within normal limits.⁷ This finding substantiated the final diagnosis. It is worth emphasising that in the present case, thorough history and meticulous examination along with detailed investigation of laboratory investigations was done.

However, definitive diagnosis requires biopsy from the offending lesion. In the present case, histopathological picture revealed congested blood vessels and mononuclear infiltrate with few osteoclasts and osteolytic area replaced with normal tissue. This finding was not typical of Gorham's disease as described in literature where neo-vascularisation and capillary proliferation are considered diagnostic features.⁸ However, it is reported in literature that histologically in the first phase there is increased vascular concentration in the bone displacing fibrous tissue with multiple anastomosing thin walled blood vessels. In the second phase there is no increased vascularisation so that in the areas which were osteolysed only fibrous tissue is replaced.⁹

Heffez *et al* produced a set a diagnostic criteria to aid diagnosis¹⁰:

1. A positive biopsy
2. Absence of cellular atypia
3. Minimal/no osteoblastic response and the absence of dystrophic calcification
4. Evidence of local, progressive osseous resorption
5. Non-expansile, non-ulcerative lesion
6. Absence of visceral involvement
7. Osteolytic radiographic pattern
8. Negative hereditary, metabolic, neoplastic, immunologic or infectious aetiology.

A majority of the above mentioned criteria were found to be compatible in the case examined by us. Hence the final diagnosis of Gorham's disease was arrived at.

Although aetiology is obscure, Gorham has explained that aetiopathogenesis may be due to proliferating capillaries from Haversian systems which cause increased vascularity in the area followed by slowing of circulation, local hypoxia and lowering of pH favouring the activity of various hydrolytic enzymes like acid phosphatase, alkaline phosphatase and lucin amino peptidase. Prognosis of the disease is dependent on which structures are involved. Lesions within the vertebral column and rib cage can potentially be debilitating or even fatal. Mandibular involvement is considered high-risk disease location as the disease can progress to involve the maxilla, skull and spine.

The natural course of disease follows¹¹:

- Active phase which shows slow progressive bone destruction
- Passive phase showing arrest of osteolysis.

Recommended treatment modalities for Gorham's disease fall into three groups¹²:

1. Surgical intervention has been suggested when feasible and involves local resection of affected bone with or without reconstruction. However, resorption of bone grafts are also reported.
2. Radiotherapy in moderate doses (40–45 Gy in 2 Gy fractions) appear to result in good clinical outcome with few long-term complications acts by accelerating sclerosis of proliferating blood vessels and prevents regrowth of these vessels.

3. Antiosteoclastic medication (biophosphonates and interferons) have proved to be most efficacious treatment with few adverse effects. Synergistic action of zoledronic acid and interferon is a powerful antiangiogenic therapy which is giving currently best results.

Learning points

- ▶ Thus, in conclusion it can be accepted that Gorham's disease is an entity associated with vague aetiology and occult progression which needs thorough investigation, meticulous treatment planning and periodic long-term follow-up.
- ▶ A high degree of clinical suspicion and awareness should be exercised while arriving at a diagnosis of Gorham's disease in the maxillofacial region.
- ▶ A wide array of differential diagnosis should be taken into consideration because diagnosis of Gorham's disease needs exclusion of other osteolytic diseases.
- ▶ Emphasis on long-term (life-long) follow-up is necessary due to progressive nature of disease and to verify spontaneous arrest and/or stabilisation of the disease.

Competing interests None.

Patient consent Obtained.

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