Case Report

Gorham's disease of mandible: A case report

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Summary: *Massive Osteolysis (MO) is a rare disease of unknown aetiology. It is even rarer in the maxillofacial region where less than 50 cases have been reported. Localised proliferation of endothelium of blood or lymphatic vessels resulting in destruction and resorption of bone is thought to be the cause of the disease. A case of MO resulting in total resorption of the left mandible in a 39 year old Nigerian male is presented. Endothelial proliferation was absent in this case, making it a variant of the established MO. It is also the first case to be reported in the West African sub region.*

Keywords: *Massive osteolysis and mandible.*

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Received for publication January 2008
Accepted after peer review June 2008
Available online June 2008 at www.jmosi.com

Introduction

Massive Osteolysis (MO) is a rare disease of unknown aetiology[1-4] that is characterized by spontaneous, progressive, and often total disappearance of bone[4]. The bone destroyed by the osteolysis does not regenerate or repair but is replaced by dense fibrous connective tissue with ultimate disappearance of the bone[5,6].

The precise aetiology of MO is poorly understood and largely unknown; however, pre existing metabolic, endocrine, malignant, neuropathic or infectious disorders have been linked with the disease[3,7]. Also reported in association with MO are increased osteoclastic activities and increased levels of Interleukin 6 (IL6) as a potential humoral mediator[5,7]. Minimal trauma has been a feature in most of the cases reported, however, it has been shown that trauma has no greater significance than that of revealing a disease already in progress[8].

MO was first described by Jackson in 1838 as a case of a boneless arm in a 12 year old male patient with complete osteolysis of the humerus[9]. Since then the disease has been reported in almost all parts of the skeleton[4], including the mandible and has been reported under various names as disappearing bone disease, phantom bone disease, vanishing bone disease, and Gorham's disease[3,6-11]. Involvement of almost every bone has been reported, although there is a predilection for bones that develop by intra membranous ossification, with shoulder girdle and mandible being the most common bones affected[12]. The first case involving the mandible was reported in 1924 by Romer in a 31 year old female patient with complete resorption of the left vertical ramus[8]. There are no fewer than 50 reported cases involving the maxillofacial region in literature.

Therefore, a rare case of massive osteolysis involving the left half of the mandible in a 39 year old Nigerian male patient is presented.

Case Report

A 39 year old Nigerian male patient domiciled in Liberia presented at the Oral and Maxillofacial Surgery clinic of the Lagos University Teaching Hospital (LUTH) in May 2005 with a complaint of deviation of his jaw and pain of about 1 year duration. Past dental history revealed he had an extraction of left mandibular wisdom tooth a year earlier because of toothache. During the extraction the left mandible fractured and was immobilized by mandibulo-maxillary fixation (MMF) for a period of 6 weeks. The patient however complained that after the removal of the MMF the pain on the left mandible persisted and he also noticed the deviation which was the cheif complaint of the patient.

Clinical examination revealed a healthy man with an obvious facial asymmetry due to flattening of face on the left side and there was a shift towards the same side and hollowing of the ramus and condylar region (Fig. 1a and 1b). The left side also appeared to be sunken with a prominence of the zygomatic bone (Fig. 1b). There was no paraesthesia or motor nerve deficiency. The left submandibular lymph nodes were firm and non tender.

Intraoral examination revealed the full complement of the teeth, except the lower left third molar that had previously been extracted. The teeth on the lower left quadrant were lingually displaced and grossly mobile.

Radiographic examination revealed complete loss of the entire left mandible, including the condylar and coronoid processes on the left side (Fig. 2a). The teeth in the left quadrant were floating due to loss of bone support (Fig. 2b). The osteolysis involved the left half of the mandible and also extended to the canine area of the right side. Routine laboratory parameters were all within normal range. Histology report of the biopsy taken from the affected side revealed fibrous cellular connective tissue, a few vascular channels and areas of haemorrhage (Fig. 3). The patient however, returned to his domicile before treatment could be instituted.

Discussion

Osteolysis of facial bones is usually as

a result of specific pathologic processes including infections, cysts, tumours, endocrine dysfunctions, metabolic disorder and immunological abnormalities[3]. However, MO is quite distinct and a rare type of osteolysis characterized by proliferation of vascular channels that results in partial or complete disappearance of one or more bones[3,13-16]. The bone destroyed by the osteolysis does not regenerate or repair but is replaced by dense fibrous connective tissue with ultimate disappearance of the bone[5]. Involvement of almost all bones in the body have been reported, although, there is a predilection for bones that develop by intramembraneous ossification with the shoulder girdle and the mandible being the most common bones affected[12]. The first case of MO was reported, involving the mandible with complete resorption of the left vertical ramus was reported by Romer[8]. Since then, there have been more than 47 reported cases involving the maxillofacial region. Only 22 cases involving the mandible alone had been reported by 2002[5,17]. Our report adds to the list of cases involving the mandible alone. The diagnosis of the present case was based on the following diagnostic criteria as suggested by Heffez et al:[18] 1) a positive biopsy for angiomatous tissue, 2) absence of cellular atypia, 3) minimal or no osteoblastic response and 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, and 8) negative hereditary metabolic, neoplastic, immunologic, or infectious aetiology. Though the disease may be limited to a single bone, polystotic cases have been reported[7]. It can also extend to the joints to affect an entire anatomic area[3]. The progression of MO from the entire mandible to the maxilla, cranium, and spine has also been reported[7].

Massive Osteolysis predominantly affects children and young adults of either sexes[5]. Most cases occur before the age of 40 years; although it has been reported in patients from 18 months to 72 years of age[17]. The present case is in a 39-year-old male; though the disease could have started at an earlier age considering the extent of the osteolysis.

MO may be asymptomatic until discovered on routine radiograph or after some form of injury often due to pathologic fracture of the involved bone[3]. Though the role of trauma as an aetiologic factor has been questioned, it is generally believed that it doesn't play any significant role[8]. It only complicates an existing disease and reveals its presence[8]. Majority of the reported cases have occurred subsequent to trauma; thus some authors have implicated this as an initiating factor[5,15,16]. The patient reported presented a history of extraction of lower left wisdom tooth because of pain. During the extraction the left angle of the mandible fractured. It is not unlikely that the mandible fractured as a result of weakness from pre-existing MO. After the release of the MMF, the pain persisted and it was then that he noticed the jaw deviation to the opposite side. Jaw deviation made him to seek treatment. The trauma therefore revealed the presence of MO in this patient. Pain may or may not be a prominent feature of MO[8,13].

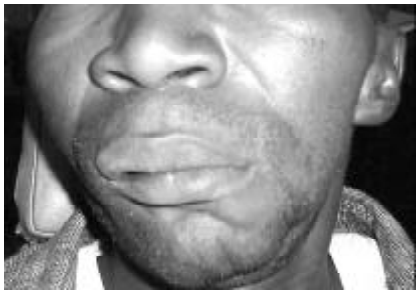

Fig. 1a: Patient at presentation (PA view) with flattening of face on the left side and shifting to the same side.

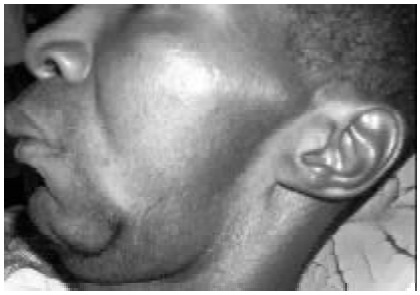

Fig. 1b: Same patient (lateral view) with prominence of the left zygomatic bone and hollowing of the ramus and condylar region.

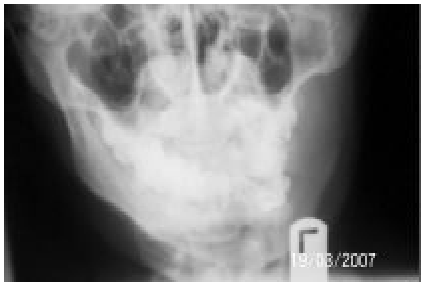

Fig. 2a: Postero-anterior radiograph showing complete loss of the entire left mandible, including the condylar and coronoid processes on the left side.

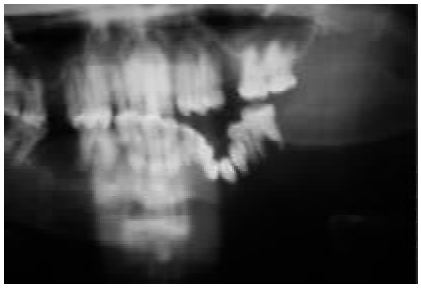

Fig. 2b: Orthopanthomogram showing osteolysis of the left mandible with floating of teeth due to loss of bone support

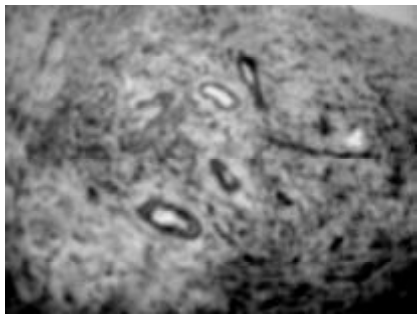

Fig. 3: Section shows fibrocellular connective tissue within which are plump and angular shaped fibroblasts and vascular channels (H & E, magnification X 100).

Radiographically, MO progresses through four stages[19]. The earliest radiographic presentation is the presence of one or several clustered intramedullary and subcortical radioluscent foci of varying sizes[8] or regions of hypodensity within distinct margins or thin radiopaque borders[5,17]. This stage resembles patchy osteoporosis[13,19]. As the disease progresses these foci undergo enlargement and then tend to coalesce[8]. Next, the cortex disappears locally as a result of either contained resorption or pathologic fracture[8,19]. Finally there is shrinkage of the ends of the affected bones and its final disappearance with tapering or porosity of the remaining osseous tissue[5,8,13]. Pathologic fracture may occur at any stage while bone regeneration is rarely seen[8,19].

The patient presented during the final stage of the disease with complete disappearance of the left mandible with the coronoid and condylar processes; and tapering of the remaining part of the right mandible. The standing teeth on the left quadrant were floating due to loss of bone support (Fig. 2b). In the early stages, it is difficult to differentiate MO from other resorptive processes. It is therefore important that a thorough history and meticulous physical examination be undertaken. Appropriate blood tests and radiographs should be requested to rule out

other common causes of osteolysis. The diagnosis should be suspected after excluding osteolytic lesions. Therefore the clinician should have a very high suspicion when other cases of osteolysis have been eliminated by physical and radiologic examination; and other appropriate blood tests.

The pathogenesis of MO is poorly understood and there appears to be no genetic basis for transmission[5]. However, it is generally believed that the pathologic process is the replacement of normal bone by an aggressively expanding non-neoplastic vascular tissue similar to haemangioma or lymphangioma[13]. This is supported by various descriptive terms by different authors in reporting the histologic changes seen in MO such as presence of haemangiomatous granulation tissue[2], lymphangiomatosis[19], numerous congested capillaries[20], haemangioedomatoma[3], irregular abnormal vascular formation[21], excessively vascular bone[5], and marrow replaced by fibrous tissue[8]. In a review of the biopsy report of 16 cases, Gorham et al[20] reported the vascular hyperplasia was of blood vessel origin in all cases and there were no malignant angiomatous changes. Fibrous cellular connective tissue and a few vascular channels, and areas of haemorrhage were histologic features seen in the present case.

Some authors have reported that osteoclastic antigens play a role in bone resorption in patients with MO[12,13], whereas others[5,7] believe that the role of osteoclasts is unclear. Gorham and Stout[3] reported the absence of osteoclasts in their series, but other authors have reported their presence[21,22]. Delvin et al[7] have suggested that bone resorption in patients with MO is due to enhanced osteoclasts activity, and that interleukin-6 (IL-6) may play a role in the increased resorption of bone.

Due to the rarity of MO, there is no standard treatment[13]. However, various treatment modalities have been employed with variable success. These include surgical and non-surgical modalities. The principal treatment modalities are surgery and radiation therapy. Surgical option includes resection of the lesion and reconstruction with bone graft[13]. However, success rate of this form of treatment is low because the bone graft is said to undergo dissolution.[23] Reconstruction should therefore be done after 12-18 months period of careful clinical and radiological observation[23]. Motamedi et al[5] suggested that surgery may eradicate the lesion when it is small.

Radiation therapy has been used with variable success in some cases when surgical resection has failed to stop progress of the disease; and it has been suggested that it presents a good clinical outcome with minimal long term complication when used in doses of 40-45Gy in 1.8 to 2Gy per fraction[24]. Advantages of radiation therapy include early arrest of endothelial proliferation and thereby limit the spread of the disease[19]. It is also good for poor surgical patients. Also, there have been reports of recalcification of affected bone following radiation therapy, a phenomenon that is exceedingly rare with other treatment modalities[13]. The major disadvantage of radiation therapy is the possibility of acute and long term side effects that vary by treatment site[13].

Other treatment modalities that have been employed are anti-osteoclastic medication (bisphosphonates) and alpha-2b interferon[13]. However, the result of these therapies is difficult to evaluate, because the disease may spontaneously arrest in some patients[18]. Although, our patient defaulted before treatment could be instituted, early detection and prompt treatment of the disease have been reported to lead to variable success and halting of the disease process.

The prognosis of the disease is variable and unpredictable[5,8,13,19]. If left untreated, MO commonly progresses to total destruction of the involved bone and possibly progression to adjacent bones[16,23]. While the disease can undergo spontaneous regression in some individuals, it can progress in some cases leading to involvement of vital structures causing morbidity and mortality[17,18].

Conclusion

A rare case of massive osteolysis involving the left half of the mandible in a 39-year-old Nigerian male is presented. The pathogenesis of MO is poorly understood and there appears to be no genetic basis for transmission, hence the prevention of progression is a main concern in MO because progression from the mandible to the maxilla, skull, and spine has been reported. For all patients who present with skeletal osteolysis, a thorough history and meticulous examination in addition to appropriate blood tests be carried out to rule out other common (in contrast to the extremely rare MO) underlying causes of osteolysis.

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Source of Support: Nil, **Conflict of interest:** None declared.