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ORIGINAL ARTICLE

Extensive osteolytic disease in the mandible of a pediatric patient: Case report

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Abstract:

Introduction: Gorham's disease, correspond to a massive osteolytic disease of the bones, characterized by destruction and resorption of one or more bones, spontaneous and progressive. The etiology of the disease is still unknown. The destroyed bone does not have the ability to regenerate or repair and is replaced by dense fibrous tissue. Bones of the skull and pelvis are the most commonly affected and in maxillofacial bones, the mandible is particularly affected. **Case report:** This study reports the case of an 11-year-old female, referred to the Oral and Maxillofacial Surgery service showing extensive area of spontaneous bone resorption, in which only the alveolar portion of the mandibular symphysis and the head of the left jaw were present. The history and clinical features observed indicated Gorham's disease. **Final considerations:** When in progressive stages Gorham's disease is difficult to be diagnosed by histopathological evaluation, due to limited remaining bone quantity, leaving only clinical and complementary exams to make allowances. The treatment is controversial in the literature. Therapy is individualized in accordance with the severity of the patient's condition and the site of involvement.

Keywords: Bone Diseases; Pathology; Bone Resorption; Osteolysis, Essential.

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INTRODUCTION

Gorham's disease (GD) is a rare bone disease also known as phantom bone disease, progressive osteolysis, disappearing bone disease, vanishing bone disease, acute absorption of bone and idiopathic massive osteolysis. As its various names suggest, involves spontaneous and progressive destruction and resorption of one or more bones[1,2].

The exact etiology of the condition is still unknown, without evidence of metabolic or endocrine explanation. Many authors have implicated trauma as the initiating factor in the majority of reported cases[3]. Destroyed bone lose the ability to repair and are replaced by fibrous tissue[2].

Histological features involves osteolysis process associated with an angiomatosis with abnormal proliferation of blood vessels and sometimes lymphatics, which could be responsible for the whole destructive mechanism. Production of local, intramedullary, lymphangiomatous endothelial tissue is in authority for the pathological change. And characteristic the start of spontaneous osteolysis is characteristic[4].

All skeleton bones can be affected, although there is a preference for humerus, axial skeleton, and pelvis. In the maxillofacial region the mandible is the most affected bone, where basal and alveolar bones are primarily affected and the ramus and the condyles are successively[1].

This paper aims to report a case of a pediatric patient with extensive involvement of massive osteolysis of the mandible, with a presumed diagnosis of GD. Due to the limitations of the presented case, it is known that the chosen therapy will be a challenge for the maxillofacial surgeon.

CASE REPORT

Eleven-year-old girl was referred to Oral and Maxillofacial Surgery division with a history of progressive mandibular bone loss. Patient and progenitor denied painful symptoms, prior trauma, or local infections as possible etiologic factors for mandibular bone resorption, and mentioned beginning of the resorption from the age of two. General and systemic examination of the patient were within normal limits.

There was acceptance of the patient's responsible in relation to the authorization for photographic records and subsequent publication of the case in scientific literature by signing the instrument of Informed Consent.

Extraoral physical examination revealed midface deficiency, with a marked flattening of the malar region and excessive sclera exposure (Figure 1). During manipulation of the lower third of face, atypical mobility was noted in the mentual region, palpation revealed the total absence of the inferior mandibular margin and the right mandibular condyle. Maximum mouth opening width was 30 mm and movement of the condylar processes was asymmetrical on opening and closing.

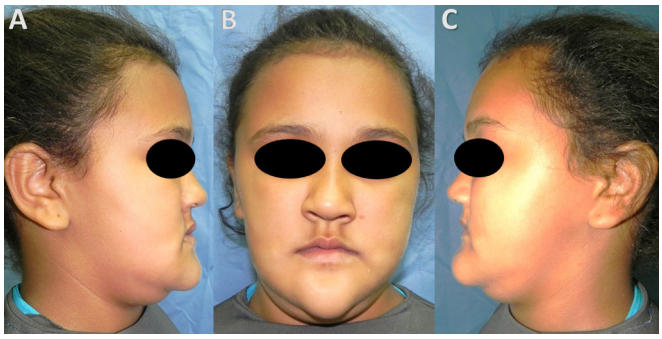

Figure 1. Lateral (A, C) and frontal (B) photos of the patient, showing a deficiency of the middle third of the face.

Intraoral clinical examination showed patient in mixed dentition stage, asymmetry between jaws resulting in intermaxillary midline deviation. Edentulous areas in the right and left posterior side of the mandible where only lower incisors remained present (Figure 2).

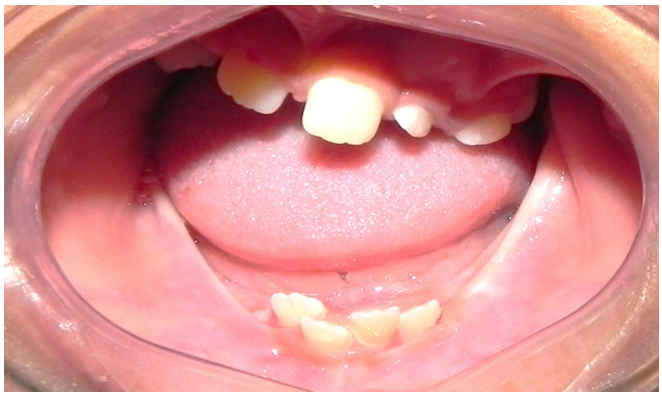

Figure 2. Intraoral photo showing intermaxillary midline deviation, edentulous areas in the right and left posterior side of the mandible. Only lower incisors remained present.

Evaluation of current and previous radiographs and tomography showed the presence of extensive progressive osteolytic processes that affected jaw almost completely, leaving only the alveolar portion of the mandibular symphysis and the head of the left jaw presented (Figure 3).

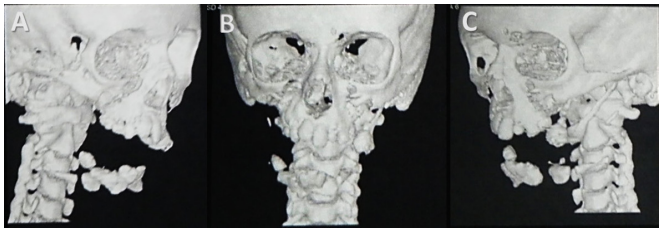

Figure 3. Computed tomography showing extensive osteolytic processes that affected jaw almost completely, leaving only the alveolar portion of the mandibular symphysis and the head of the left jaw presented. (A) Right, (B) front and (C) left view.

Possibility of incisional biopsy of the alveolar osteolytic region was discarded due to the small amount of remaining bone that the patient presented. According to the standard bone resorption described and presented by the patient, diagnosis for Gorham's Disease was presumed.

DISCUSSION

The pathogenesis of Gorham's disease remains unclear but is believed to be related to vascular proliferation. Despite Zhong et al.[5] report that Gorham's disease affects mostly young adults with no significant gender difference, patient was still pediatric, having not yet reached the stage of puberty, corroborating with Kiran & Anupama[6] who reported that most cases occur in children or adults aged less than 40 years.

Although the main signs and symptoms indicators of GD are intense pain, localized swelling and pathological fractures[4,7] the patient described showed none of those symptoms, corroborating with Kiran & Anupama[6] that report the process is painless. Those authors[6] described the evolution of disease as suddenly start, rapid progress, and then, bone is replaced by a thin layer of fibrous tissue surrounding a cavity.

Corroborating with the literature[1] that says men or women from any age group may be involved, and that most cases are discovered before the age of 40 year the reported patient was 11 years old, and progenitor mentioned beginning of the resorption from the age of two. No familial predisposition has been found.

Huang et al.[7] felt difficulty in diagnosing the disease because it had non-specific presentation in both physical signs and histopathology. Therefore their diagnosis was by exclusion. When neoplastic, metabolic, infectious, hereditary, and immunologic disorders are ruled out, the diagnosis of GD must be considered.

We also felt difficulty in diagnosing Gorham's disease, since in the described case the biopsy was contraindicated, and agree with the authors[7], who report that despite absence of bone histopathology, considering the history and clinical manifestations, the pathological findings of the soft tissues, the biochemical and haematological results, the diagnosis of GD in the case presented is reasonable and logical.

Al-Jamali et al.[2] presented a case report where the histopathologic and examinations showed mainly proliferating lymphatic tissues with evident dilated lymphatics, which confirm the role of disordered lymphangiogenesis in the pathogenic of GD. In the present case there was no possibility to perform biopsy because of the extensive bone resorption, which was a limitation.

Challenge in this disease lies in both: how to diagnose and how to treat[8]. GD treatment is controversial in the literature, given the rarity of this disease entity, there is no standard therapy available. Hammer et al.[9] say that this is attributable to the rarity of the disease and to the lack of understanding of its underlying pathogenetic mechanisms. Therapy is individualized in accordance with the severity of the patient's condition and the site of involvement.

The use of bisphosphonates for stabilization of bone resorption has been discussed in the literature. Hammer et al.[9] noticed a stable clinical picture in a 45-year-old male, treated with pamidronate 30 mg IV every 3 months, during 2 years. This treatment protocol did not show any progression of bone destruction of patient's ribs, according with radiological evidence.

On his literature review, Patel[10] concludes that most patients have been treated with surgery and/or radiation therapy[8,10,11]. Which corroborates with Tateda et al.[12], that combined conservative and surgical treatments for a 15-year-old patient with massive osteolytic lesions in cervical spine, from C1 to C5. Authors started treatment with chemotherapy using interferon α-2b (1,000,000-4,000,000 units/day) and pamidronate disodium (30 mg/week) for about one and a half years, until the osteolytic condition regressed, and then a 36 Gy radiotherapy was performed. One year after radiotherapy, no progression of osteolysis was noticed, so spinal fusion from C2 to C5 was planned to prevent further progression of kyphotic change.

Kim et al.[13] associated surgical treatment with the use of bisphosphonates and radiotherapy. The authors were successful in treating a humeral pathological fracture of an 18-year-old diagnosed with Gorham's disease who underwent autologous vascularized fibular

graft. After 2 weeks postsurgery, intravenous zoledronic acid infusion was started once a month at a dose of 4 mg for a duration of 6 months. Radiation therapy was started at a single dose of 2.0 Gy (20 cycles) for the duration of 4 weeks with a total dose of 40 Gy. Radiographs of the patient 7 years after surgery and 10 years after the first surgery showed evidence of bone union and no signs of osteolysis

Rauh & Gross[14] speculate that watchful waiting, especially in children, might be an alternative option, but requires careful monitoring. As therapeutic planning for the case described, bone graft and reconstruction with prosthetic individualized devices has been considered.

FINAL CONSIDERATIONS

GD is a disease that when in progressive stages is difficult to be diagnosed by histopathological evaluation, due to limited remaining bone quantity, leaving only the clinical and imaging exams to make allowances. Treatment for this disease is also quite controversial in the literature and represents a challenge to the maxillofacial surgeon.

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