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CASE REPORT

Gorham stout syndrome in maxilla and femur

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Abstract:

Gorham Stout syndrome (SGS) is a rare disease that affects the bones causing idiopathic osteolysis. The prognosis of SGS is quite variable and impossible to predict, in most cases, bone destruction progresses from months to years, the affected area does not regenerate or repair itself. The etiology of the disease is unknown. The aim of the present study is to report a case in female patient showing a lesion in the left jaw compatible with the SGS. A 45-year-old female patient, was referred to the CTBMF team. After performing imaging tests to perform the differential diagnosis, they suggested as differential diagnosis of maxillary ossifying fibroma lesion and fibrous dysplasia, in relation to femoral lesion, was suggestive and enchondroma. The proposed treatment was surgical removal with curettage of the lesion in the femur and maxilla. With the result of the histopathological examination, the diagnosis of SGS was concluded. During the postoperative follow-up period, the lesion showed no signs of relapse, guaranteeing success in the treatment.

Keywords: Osteolysis; Maxilla; Neoplasms; Diagnosis.

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INTRODUCTION

Gorham Stout syndrome (SGS) is a rare disease that affects the bones causing idiopathic osteolysis. In place of the bone matrix there is deposition of tissue that can be connective, blood vessels and lymphatic. The etiology of the disease is still unknown. In most cases it involves only one bone, the most recurrent are the pelvis, humerus and mandible. About 50% of all patients report an episode of trauma prior to diagnosis, while maxillofacial involvement is observed in approximately 30% of the patients, and simultaneous involvement of the maxilla and mandible can occur[1].

Radiographically the initial changes consist of radiotransparent intramedullary foci, of variable size, with indefinite margins. Its histopathological features in the early stages consist of a non-specific vascular proliferation interposed with fibrous connective tissue and a chronic inflammatory infiltrate of lymphocytes and plasma cells in the late stages the tissue from the areas of bone loss is more collagenated, and no evidence of repair by the formation of new bone[2].

In 1955, Gorham and Stout reported the main pathological features of what was termed "missing bone disease with intraosseous vascular alterations". The differential diagnosis of Gorham's syndrome includes bone hemangioma, angiosarcoma, essential osteolysis and hereditary osteolysis. The radiographic pattern is very similar, but in these diseases there are no large pleural effusions and other pulmonary alterations[3].

The prognosis of SGS is quite variable and impossible to predict, in most cases, bone destruction progresses from months to years, resulting in total loss of bone or affected bones. The affected area does not regenerate or repair itself. The signs and symptoms of this pathology include dental mobility, pain, malocclusion, mandibular deviation and facial asymmetry[4].

The objective of the present study is to report a case in female patient presenting simultaneous lesion in the jaw and femur of the left side, with clinical suspicion of Gorham Stout Syndrome.

CASE REPORT

A 45-year-old female patient, leucoderma, former smoker, referred to the Maxillofacial Surgery and Traumatology (CTBMF) team. The patient complained of spontaneous and progressive mobility of the dental elements 24 and 25 and increased volume in the infraorbital region for approximately six months. It contained a panoramic radiograph demonstrating extensive apical lesion in said teeth. In the clinical examination, bulging was observed in the left infraorbital region and left maxillary tuberosity, grade III mobility of the dental element 25 with mucosa without alteration of color and consistency. Both teeth were vital as tested for vitality.

At first, an incisional biopsy was performed and the lesion was submitted to histopathological examination, which presented nonspecific vascular proliferation interspersed by fibrous connective tissue, absence of malignancy (figure 1 and figure 2).

From the performed knee and face examinations the suspicion of SGS arose, then a scintigraphic examination was performed which can be observed an important area of increase of non-specific metabolic activity in the maxilla and in the distal epiphysis of the left femur (figure 3 and 4).

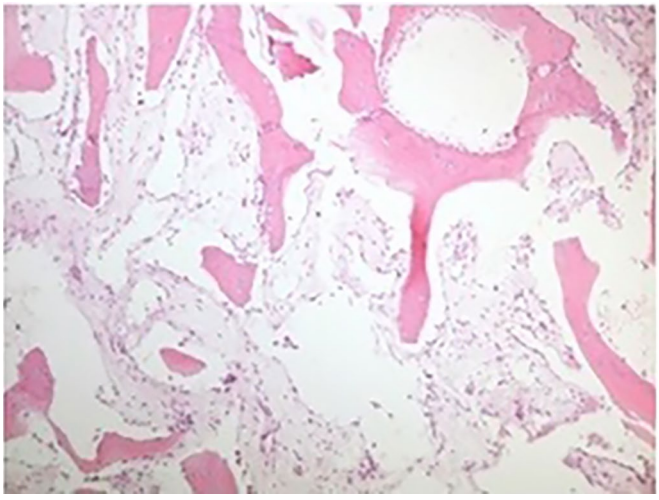

Figure 1. Demonstrating nonspecific vascular proliferation interspersed by fibrous connective tissue. HE staining 10x magnification.

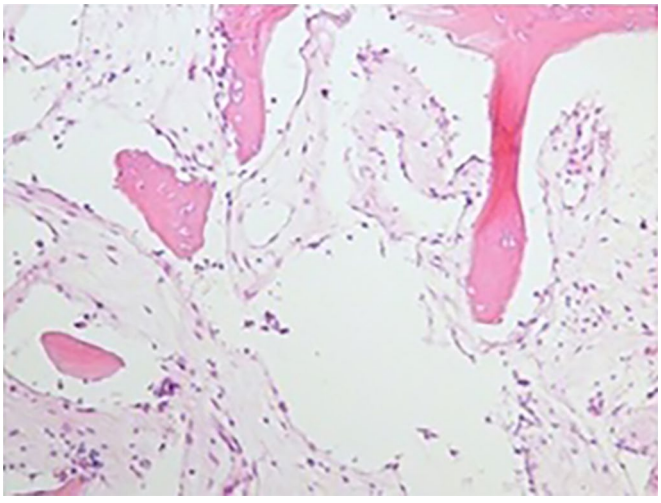

Figure 2. Demonstrating nonspecific vascular proliferation interspersed by fibrous connective tissue. HE staining 40x magnification.

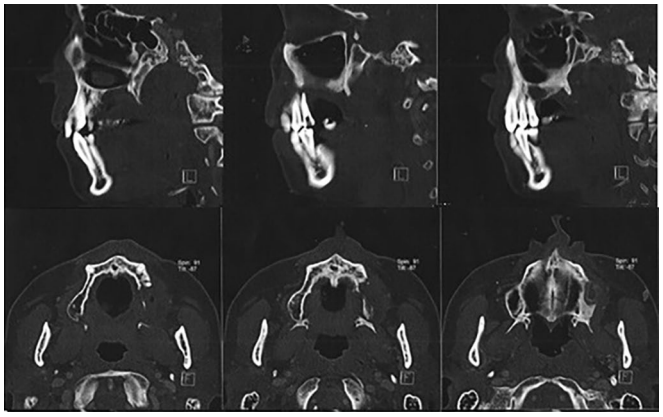

Figure 3. Scintigraphy face axial demonstrating area of increase of non-specific metabolic activity in the maxilla on the left side.

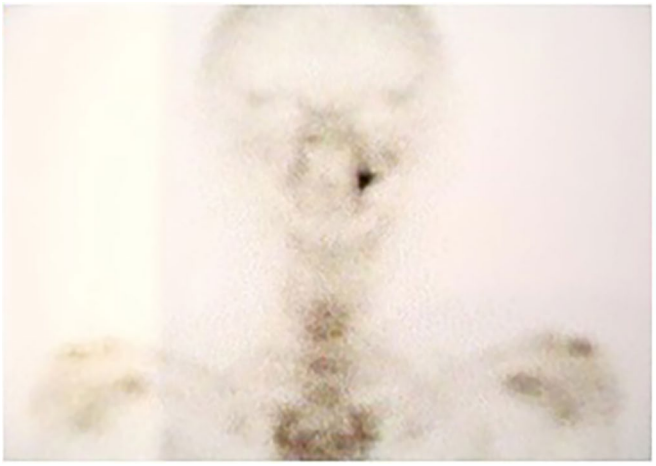

Figure 4. Scintigraphy axial knee demonstrating area of increase of non-specific metabolic activity in the left femur.

An x-ray of the left knee was performed showing expansive lesion in the distal third of the femur measuring 4.5cm in its largest diameter, with well-defined contours that suggested a diagnostic hypothesis of osteochondroma. Thus, magnetic resonance imaging (MRI) was used to demonstrate the presence of an expansive lesion with benign features in the distal portion of the femur. With the hypothesis of SGS without apparent etiology, incisional biopsy in the femur was performed and adding the imaging exams the differential diagnosis was compatible with enchondroma.

In this way, a computerized tomographic examination of the face was performed (figure 5), which demonstrated rupture of the floor of the maxillary sinus on the left side and destruction of the alveolar bone in the region of the dental elements 24 and 25, which did not present root resorption. Tomography combined with clinical examination suggested a diagnostic hypothesis of ossifying fibroma and fibrous dysplasia. The patient contained several septic foci and multiple extractions

Figure 5. Computed tomography of the sagittal face and axial demonstrating osteolytic area in the region of elements 24 and 25. As well as its relation with neighboring structures such as the maxillary sinus.

were performed. The patient was referred for endodontic treatment to preserve the teeth involved in the lesion.

When discarding the hypotheses of Fibroma Ossificante (FO) and Fibrous Dysplasia (FD), together with the patient's clinical history, exams and anatomopathological characteristics, surgical removal of the lesion on the femur and maxilla was proposed preserving the dental element 24. The lesion presented a clinical appearance of mild consistency and with a good vascularization and absence of a fibrous capsule (figure 6 and 7).

By adding the clinical, imaging and histopathological data, the diagnosis of Gorham Stout Syndrome can be concluded. The patient remained under clinical and radiographic monitoring. After eighteen months, there was no recurrence of the lesion showing the success of the surgical treatment (figure 8).

DISCUSSION

SGS affects bones causing osteolysis, macrophages play a key role in this process, as they activate the osteoclasts, secrete enzymes such as interleukin-6 and tumor necrosis factor that stimulate bone resorption[5].

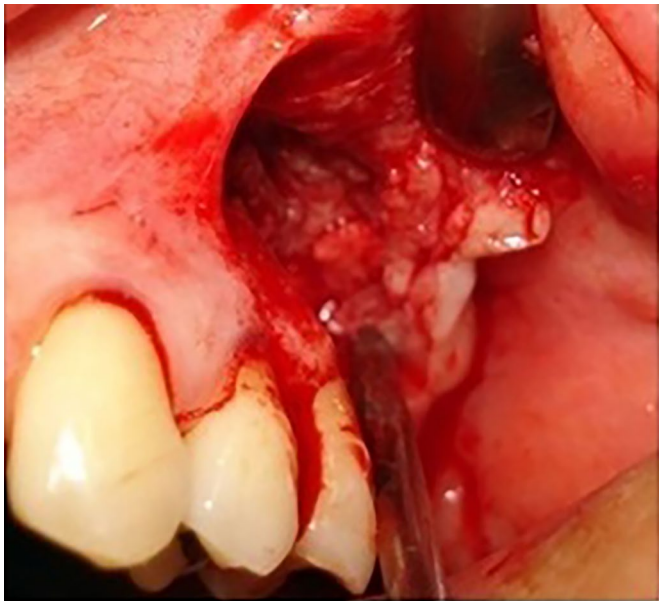

Figure 6. Transoperative removal of the lesion.

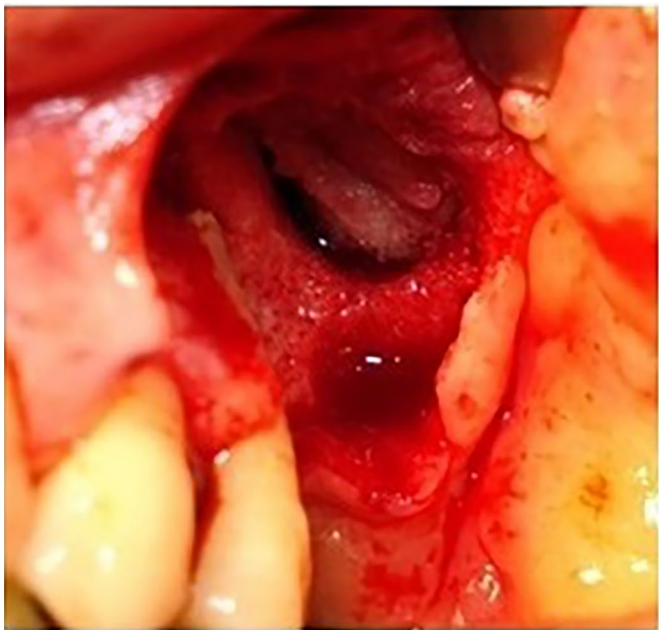

Figure 7. Transoperative after removal of the lesion.

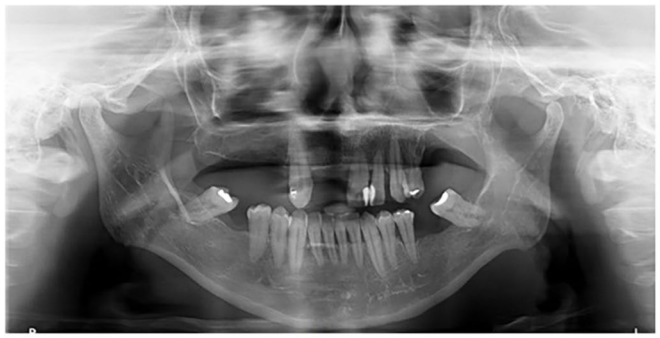

Figure 8. Panoramic radiography after 1 and a half years of follow-up with no recurrence of the lesion.

In the lesion, inhibition of osteoblasts prevents bone neoformation, macrophages also synthesize epithelial growth factors such as VEGF-A, which stimulates vascular and lymphatic vessels to proliferate at the site of osteolysis, thus forming a tissue that gives rise to the name of the lesion such as massive osteolysis[6,7]. The etiology of the disease is unknown. However, it is believed that some cases may have involvement with previous trauma[7,8].

The diagnosis of SGS is given after ruling out all the differences diagnoses, since it is a rare syndrome. Benign lesion like FO is part of the differential diagnosis. It proliferates fibrous connective tissue and may contain bone or cementum, and thus is considered a fibro-osseous lesion[4]. When it occurs in the maxilla there is predilection for the canine fossa region and zygomatic arch area. It can also cause mobility in the teeth in which they are affected and increase in volume in the area involved[8]. In histopathological examination there is similarity to SGS, because there is a fibrous connective tissue and bone fragments. However, on radiographic examination SGS is radiolucent with indefinite margins, whereas FO has a mixed image and defined margins[8,9].

Fibrous dysplasia was also a differential diagnosis. It is a benign lesion that can affect only one bone called a monostotic, or involve more than one bone called polyostotic, is usually unilateral[10]. It is congenital, it replaces normal bone with fibrous connective tissue[11]. Histopathological examination was similar to SGS by the fibrous connective tissue composition and the presence of hyperactivity of osteoclasts; however, in fibrous dysplasia, the lesion tissue image is Chinese character format, which was not identified in the histopathological examination of the patient.

In histopathological examinations of the femur the differential diagnosis was of enchondroma and osteochondroma. Osteochondroma is a benign lesion that normally affects the metaphysis of long bones, of unknown aetiology, causes painless bone growth with a radiopaque radiographic image[12]. The enchondroma, benign tumor, has a radiographic image similar to the osteochondroma and causes proliferation of hyaline cartilage in the bones that it affects. However, in SGS there is a bone destruction and the radiographic image appears in a radiolucent way, thus differentiating it from the diagnostic hypotheses[12,13].

The prognosis of the lesion depends on the location, extent and size of the lesion. In this case report, the jaw is more affected than the maxilla. The anatomical structures present therein imply greater care for the treatment and impediment of lesion growth[14].

The maxillary sinus, close to elements 24 and 25, was affected by the rupture of the floor. This was also seen in a case report in which SGS was located in the region of teeth 25 and 26 and caused cortical reabsorption of the maxillary sinus by 1.7cm[14]. This reabsorption may impair the functions of the maxillary sinus as air heating, develop infections and aesthetic deformities if the lesion progresses. If the lesion is not treated, total resorption of the affected bone may occur and, in addition to causing aesthetic complications, affect the patient's quality of life, making speech and chewing difficult[5,15].

When diagnosing the lesion as SGS, the proposed treatment was surgical removal with local bone curettage. There are also studies carried out with conservative treatments and with the use of adjuvant treatments to aid in the stabilization of the lesion[1,7]. Although difficult to diagnose, the rapid decision to surgically remove the lesion may prevent the spread of the lesion to other bones, preventing other lesions from occurring concomitantly to the syndrome[15].

The coadjuvant treatment with the use of the beta blocker propranolol has been proposed. As propranolol acts to inhibit endothelial growth factor (VEGF-A), a study with a patient with SGS has shown that when administering 2mg per kg of propranolol per day to the patient, reduction of the growth factor of the lesion occurs and thus stabilizes it[7]. Therefore, the use of drugs for the control of the lesion causes in a long time of treatment when compared with the surgical one[7].

In some cases delayed removal of the lesion may result in other lesions resulting from the syndrome that may result in death[5]. In one case of SGS in the chin region of the mandible, the patient was submitted to the use of bisphosphonate pamidronate 30 mg once a month for 21 months. After this period, the mandible, hyoid bone and mastoid process were completely reabsorbed and the patient's death subsequently occurred due to the complications of these bone resorptions[5].

Treatment of the disease is a challenge, and can be accomplished with the use of drugs such as bisphosphonates and interferon, with radiotherapy or with surgical resection[9,16]. Bisphosphonates reduce bone resorption in a dose-dependent manner, mainly by inhibiting recruitment and promoting apoptosis of osteoclasts, but in several reports in the literature a strong relationship has been identified between the chronic use of these drugs and a serious side effect of therapy, that is a osteonecrosis of the jaws[16]. In this report, surgical removal with local bone curettage was successful after one year of follow-up. Thus, the prognosis will vary according to the characteristics of the lesion, its location and which treatment chosen by the surgeon.

CONCLUSION

The SGS has a difficult diagnosis that must be reconciled with different examinations such as computed tomography and histopathological examination. From the discards of the differences diagnoses can reach the conclusion of the diagnosis. In this case report, the decision in the treatment decision can allow a better quality of life for the patient. In this way, the lesion did not recur after follow-up, guaranteeing successful treatment.

There was no conflict of interest between the authors.

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