Gorham-Stout Disease: Vanishing Bone Disease

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**Abstract**

Gorham-Stout disease is an extremely rare non-hereditary condition in which spontaneous, progressive resorption of bone occurs. It has no age, sex or racial predisposition. Because of its low incidence and variability in clinical presentation, the diagnosis is apparently missed. Moreover, there is no protocol or specific guidelines for the treatment. We present the data of patients less than 27-year-old, diagnosed with Gorham-Stout syndrome out of which most of them are with involvement of the left mandible, and discussed symptoms, diagnostic and therapeutic features. Diagnosis of this disease is critical and requires extensive workup and investigations with a combination of clinical, blood tests, radiological and histopathological findings. Surgical biopsy with histological identification of the vascular or lymphatic proliferation within the affected bone is an essential. As it has serious morbidity, it should be ruled out in every osteolytic bone condition. The first case of GSD in maxillofacial region was reported by Romer in 1924, Handbuch der speziellen pathologischen Anatomie and histology, Springer, Berlin. Since then, there have been few case reports and reviews of the same maxillofacial region. We present cases of GSD in maxillofacial region reported in literature along with our experience of cases.

**Keywords:** Gorham-Stout Syndrome; Acute Osteolysis; Mandible; Diagnosis; Antivirals Use

**Introduction**

Gorham-Stout disease (GSD), also referred as vanishing bone disease, massive osteolysis, and more than a half-dozen other terms in the medical literature, is a rare bone disorder characterized by progressive osteolysis in conjunction with the overgrowth (proliferation) of lymphatics. Patients affected with GSD, experience progressive destruction and bone lysis. Multiple bones may become involved. Areas commonly affected by GSD include the ribs, spine, pelvis, skull, collarbone (clavicle) and jaw bone (maxillofacial area). Pain and swelling in the affected area can also be a symptom. Bones affected by GSD are more prone to fractures because of reduced bone mass (osteopenia). The severity of GSD can vary from one person to another and the disorder can potentially cause disfigurement and functional disability of affected areas.

Bone is continuously engaged in a process of remodeling. In individuals with GSD, affected bone is broken down, but no new bone growth occurs. Basically affected bone disintegrates and is replaced by a fibrous band of connective tissue. Osteolysis may result in progressive bone loss of all or a portion of affected bones [1].

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As the disease advances, bone deformity occurs with further loss of bone mass. In the tubular bones (the long bones of the arms and legs), a concentric shrinkage is seen which has been depicted as having a “sucked candy” appearance. Once the cortex (the outer shell) of the bone is disrupted, vascular channels may invade adjacent soft tissues and joints. Eventually, complete resorption of the bone occurs and also can extend to adjacent bones, though spontaneous arrest of bone loss has been reported on occasion. The therapeutic procedure remains controversial because of the rarity and progressive osteolysis of this disease, while reconstructive surgeries are used in certain cases in an attempt to recover the function of the involved bone. According to prior records, Gorham-Stout syndrome usually start in a single bone (or very few bones) or contiguous bones around one focus [2,3].

Complications from Gorham’s disease may occur when fluids build-up in the space between the membrane that surround each lung and line the chest cavity [1]. This can have serious consequences, including loss of protein, malnutrition, and respiratory distress and failure [9].

Methodology

This cross-sectional study was conducted from January 2015 - December 2018. Tested sample size was calculated using WHO software for sample size which turned out to be 321. Patients under 27 years of age were recruited in this study. A history and examination form designed particularly for the study was filled by concerned doctors. Records were reviewed to collect detailed information about every patient.

Patients of age ≤ 27 years regardless of gender presenting with principal complain of facial and neck pain, headache, difficulty chewing, localized symptoms in and/or around the ear (swelling, redness) associated with or without vertigo were included in the study. Patients having systemic illnesses like autoimmune disorders, metabolic syndromes and neoplastic disorders were excluded from the study.

Investigations and Results

Biochemical and haematological tests are important to exclude other diagnoses. These tests were unremarkable. Alkaline phosphatase is done which was found to be elevated in few patients with an associated fracture. Radiographic examination revealed massive bone resorption with widening of the ligament spaces of joints and the affected teeth. The histopathology reports were usually inconclusive, designating a nonspecific inflammation.

Out of 321 patients, most of the patients presented with complaints of facial pain, especially jaw pain 46%, neck pain 21%, headache 24% and 9% with no pain at all. Almost 66.87% [Relative risk (RR) = 0.33, 95%CI: 0.18] experienced difficulty chewing with (27%) or without (12%) swelling. Other symptoms were swelling in/around ear 27% and vertigo 3%. 56% of the patients used antivirals within 5 years of developing these symptoms, 31% doesn’t and 13% are unaware of usage. 21% of the patients have cutaneous manifestations related to GSD, 57% does not have any manifestations while 22% had pre-existing skin problems.

Figure 1: Gingival overgrowth and crowding.
Discussion and Conclusion

Numerous studies concerning the etiopathology and clinical presentation of Gorham-Stout syndrome have been reported, along with radiographic findings and therapeutic options, in order to raise the awareness of this rare disease.

The clinical presentation of GSD is dependable on the affected sites. Few patients have experienced abrupt onset of pain and swelling or a pathological fracture, while others have presented with a history of an insidious onset of pain, limited movements and progressive weakness in the affected area [4]. The disease is not usually accompanied by any systemic symptoms [5]. In most cases, the osteolysis may resolved spontaneously, and therefore the prognosis is generally good if vital structures are not involved.

As the disease and its relation with antivirals is so rare, no standard therapy and treatment is available. The treatment modalities include surgery, radiotherapy, etidronate therapy and the use of α-2b interferon [2,4].

A final diagnosis of Gorham-Stout syndrome is difficult. Laboratory findings are not specific and are of no value in the diagnostic procedure, except few patients have shown mild increase in alkaline phosphatase. Radiographs provide the most significant clues for obtaining a diagnosis. CT scanning and three-dimensional reconstruction are more useful for accurately assessing the extent of bone destruction at the time of diagnosis. MRI is used to define the extent of vascular formation and the involvement of the adjacent soft tissue. The histological findings depend on the phase in which the disease is diagnosed. In the first of the two phases, the bone-displacing fibrous tissue section exhibits a higher concentration of blood vessels, whereas only fibrous tissue is detected in the second phase [6,7].

Hereditary and essential osteolysis, tumours, skeletal angiomias, infection and other causes of osteolysis should all be ruled out before a differential diagnosis of Gorham-Stout is made, for which blood tests played an essential role [8].

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Bibliography

9. What is Gorham’s Disease? Lymphangiomatosis and Gorham’s Disease Alliance.

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