GORHAM'S DISEASE

Synonyms:
Gorham-Stout Syndrome, Idiopathic Massive Osteolysis, Disappearing Bone Disease, Vanishing Bone Disease, Phantom Bone Disease. (1)

Introduction:
Gorham-Stout's disease (GSD) is a rare condition of unknown etiology that results in spontaneous bone resorption. The involvement of the musculoskeletal system is characterized by progressive bone destruction and proliferation of vascular and lymphatic channels in areas adjacent to the osteolytic bone [1-3]. There are various schools of thought regarding bone resorption. Graham and Stout suggested that trauma may trigger the production of vascular granulation tissue (by stimulation of VEGF) and osteoclasts are not necessary for bone resorption [3]. However several other reports suggest that osteoclasts play a major role in the resorption. The histological samples of GSD patients have shown increased numbers of active osteoclasts in a few series/ case reports. These groups of scientists believe that IL-6 and macrophage colony-stimulating factors induce osteoclast formation [3,4,5,6].

Clinical features:
The disease can occur at any age but is usually seen in younger patients below 40 years of age. It has no predilection for gender, race, and geography, although some authors have noticed its prevalence more in males [1,3].
It can affect one or multiple bones with more frequent involvement of the maxillofacial region. The other commonly affected bones are the ribs, spine, pelvis, skull, and clavicle. Pain, swelling, and functional impairment of the affected region are the common presentations. Other orthopaedic manifestations include pathological fractures, absence of bones, bone deformities, and kyphosis [1-3].
Because of unifocal or multifocal vertebral and spinal cord involvement, the disease can lead to major neurological deficits, and other neurological complications like CSF leakage, meningitis and hearing problems. Chest involvement leads to pleural or pericardial effusion, chylothorax, or mediastinal mass. Authors have reported lymphangiomatous malformation in the cutaneous manifestation of GSD, which may present as dark plaque or cutaneous fistula.

Diagnosis

There is no specific test or procedure to diagnose GSD and hence the diagnosis is partly a diagnosis of exclusion. A diagnosis is made based upon specific characteristics symptoms, a detailed patient history, a thorough clinical evaluation and a variety of specialized tests including biopsies and specialized imaging techniques [7].

A high degree of clinical suspicion and radiological imaging is essential for diagnosing GSD. In a plain radiograph, the findings are different according to the stages of the disease. In the initial phase, subcortical and intramedullary radiolucent foci resembling patchy osteoporosis are seen. As the disease progresses, osteolysis without osteosclerosis or periosteal reaction results in bone mass loss, and concentric shrinkage of long bones of the upper and lower extremity, thus giving tubular bones a 'sucked candy' appearance. Eventually, near-complete resorption of bones occurs, justifying the name 'Vanishing bone disease' [7,8].

CT scan shows bone loss and extension to the soft tissue. The lesion appears hypointense in T1 weighted and hyperintense in T2 weighted MRI. Contrast-enhanced MRI is particularly useful to detect the reticular pattern of the substituting soft mass in place of resorbed bone. This reticular pattern is due to the vascular and/or lymphatic channels within the bone at the region of active osteolysis. In PET/CT, F-NAF turned out to be more specific and sensitive to identify osteolytic foci in comparison to F-FDG. Bone Scintigraphy shows increased uptake in vascular/ lymphatic proliferation and a decreased uptake can be observed in the osteolytic region of vanishing bone.

Bone biopsy reveals the presence of abnormal lymphatic tissue and characteristic bony changes. Hafeez et al. proposed eight histopathologic and clinical criteria that can be used for the diagnosis of GSD [9]:

(i) A positive biopsy (angiomatous tissue with abnormal lymphatic channels and numerous osteoclasts)

(ii) Absence of cellular atypia

(iii) Minimal or no osteoblastic response and absence of dystrophic calcifications

(iv) Evidence of local progressive bone resorption

(v) Non-expansive, non-ulcerative lesion

(vi) Absence of visceral involvement

(vii) Osteolytic radiographic pattern and

(viii) Negative hereditary, metabolic, neoplastic, immunologic and infectious etiology

Treatment

There is no definitive cure for this disease [7-10]. Medical management may include bisphosphonates, calcium salts, vitamin D, α-2b interferon, and sirolimus (mTOR inhibitor) [4,6,10]. Surgical management involves resection of the lesion and reconstruction with the help of a bone graft or prosthesis [7]. However, the bone grafts can undergo osteolysis if used during the active disease process. Hence, many surgeons prefer radiotherapy treatment to the lesion site to halt the process of osteolysis, so that it can be replaced by fibrous tissue. Few surgeons prefer to use medical management such as anti-resorptic medication (bisphosphonate) instead of radiation therapy to minimize morbidity. The dictum is that the bone graft should be used once there is the stabilization of the osteolytic process. Even radiation therapy can be opted for along with antiosteoclastic medications (Zoledronic acid, Pamidronate) for small lesions without fracture. Radiation therapy (total dose of 30 to 45 Gy) has also been effective in treating chylothorax, which is sometimes associated with GSD [7-8].

Conclusion

GSD is a multi-factorial one, and the etiopathogenesis of the disease has not been fully understood. The diagnosis is quite challenging, while the prognosis is poor.

References:

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IOA Clinical record format (Google questionnaires format)

(Gorham's disease)

	Preoperative	Postoperative			
		3m	6m	12m	5yrs
Demography Age Sex Occupation Address					
Clinical details Age at the time of diagnosis Pain, swelling, tenderness, fracture, ROM etc. Bone(s) involved Patient Consent taken?					
Radiographic findings (upload image) Omit patient details on images and include images after consent					
MRI/CT (upload images) Any other Imaging Omit patient details on images and include images after consent					
Histopathology					
Treatment details					
Medications (dose, duration)					
Surgery details					
Radiation therapy details					
Follow up (recurrence/complications/sec surgery etc.)					