Vanishing bone disease (Gorham's disease) – A diagnostic dilemma

Dr. Mukulchotrani*, Dr. D.S. Kulkarni**, Dr. Y. P. Sachdev***

Abstract

Vanishing bone disease (Gorham-Stout syndrome) is a rare entity of unknown etiology, characterized by destruction of osseous matrix and proliferation of vascular structures, resulting in destruction and absorption of bone. Despite the extensive investigation of the pathogenetic mechanisms of the disease, its etiology hasn't been clarified and several theories exist. The syndrome can affect one or multiple bones of the patient, including the upper and lower extremities, the spine pelvis and skull . The clinical presentation of a patient suffering from vanishing bone disease includes, pain, functional impairment and swelling of the affected region, although asymptomatic cases have been reported, as well as cases in which the diagnosis was made after a pathologic fracture. In this short review we summarize thatradiologist andclinicians should be aware of the existence of this rare entity as only right diagnosis lead to proper to therapeutic approach.

Introduction

Vanishing bone disease is a rare entity characterized by destruction of osseous matrix and proliferation of vascular structures with benign origin (1). Despite the extensive investigation of the pathogenetic mechanisms of the disease, its etiology hasn't been clarified. The first that described this entity was Jackson in 1838, who reported the case of a young man with a gradually vanishing humerus (2). Moreover, in 1955, Gorham and Stout published a paper, which correlated the massive osteolysis noted in the disease with hemangiomatosis (3), that seems to have played an important role in the fact that vanishing bone disease is also called "Gorham-Stout syndrome".

This syndrome is considered as the type IV of osteolysis, according to Hardegger et al (4), among five types: type I is hereditary multicentricosteolysis with dominant transmission, type II is hereditary multicentricosteolysis with recessive transmission, type III is nonhereditary multicentricosteolysis with nephropathy and type V is Winchester syndrome, defined as a monocentric disease of autosomal recessive inheritance (4).

Despite its previously mentioned benign character, its prognosis is unpredictable (5) and the presence of several serious complications in some cases cannot be ignored. Therefore, meticulous research has been done concerning the molecular mechanisms of the disease.

Case report

A 30 yr old female presenting with dull aching pain in left shoulder since 2 months.Pain was insidious in onset. She also complained of limitation of movement and swelling since 1month.

There was no history of trauma. There was no history of fever and weight loss. There was no significant past history related to left shoulder joint. No similar complaints in past. No history of tuberculosis or any other bone disease.

Xray of left shoulder was done.

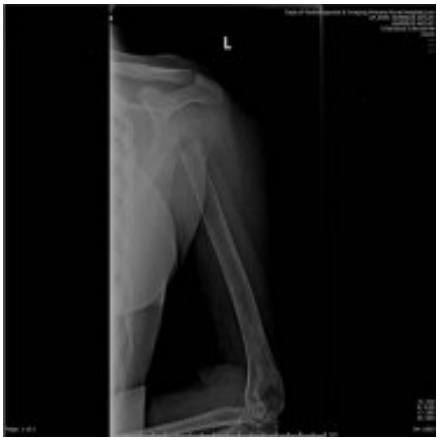
Radiograph revealed, there was profound osteolysis with resorption of head of left hmerus bone and lack of

* Resident, ** Professor *** Professor & HOD

Corresponding author :
Dr. Mukulchotrani
Department of Radiology ,
Rural Medical College of PIMS (DU), Loni. Maharshtra, India.
mukulchotrani@yahoo.co.in

compensatory osteoblastic activity or periosteal reaction. Rest of the humerus appears normal. Scapula appears normal. Glenoid appears normal.

Histopathology report was, regenerative bony trabeculae along with fibromuscular tissue, foci of ossifications are seen in the fibromuscular tissue. There is no evidence of malignancy in the section studied.



Discussion

It was first reported by Jackson in 1838, but later defined by Gorham and Stout in 1955.

Gorham disease or vanishing bone disease is a poorly understood rare skeletal condition which manifests with massive progressive osteolysis along with a proliferation of thin walled vascular channels. Other names for this condition include progressive massive osteolysis, Gorham-Stout disease and phantom bone disease.

There is no evidence of a malignant, neuropathic, or infectious component involved in the causation of this disorder. The mechanism of bone resorption is unclear.

The clinical course is generally protracted but rarely fatal, with eventual stabilization of the affected bone being the most common sequelae.

The disease starts in one bone but may spread to involve adjacent bony and soft tissue structure.

Gorham's disease is thought to be non-hereditary and there is no recognized gender predilection. It can potentially occur in any age group although most reported cases have been in young adults(7).

Pathology- The osteolysis is thought to be due an increased number of stimulated osteoclasts(8), which is likely secondary to abundant non-neoplastic vascular and lymphatic proliferation in the affected region (10). The bone is subsequently replaced by variable amounts of fibrous connective tissue that is hypervascular (11).

The pathological process is the replacement of normal bone by an aggressively expanding but non-neoplastic vascular tissue, similar to a hemangioma or lymphangioma.

In the early stage of the lesion, the bone undergoes resorption, and is replaced by hypervascular fibrous connective tissue and angiomatous tissue. Histologically, involved bones show a non-malignant proliferation of thin-walled vessels; the proliferative vessels may be capillary, sinusoidal or cavernous. In late stages, there is progressive dissolution of the bone leading to massive osteolysis, with the osseous tissue being replaced by fibrous tissue. The stimulus that generates this change in the bone is unknown (12)

Location-Gorham disease can potentially involve any bone. Reported sites include:

Humerus (first reported case), Shoulder girdle, Pelvis, Skull (7) and Mandible

Splenic lesions (cysts) and soft-tissue involvement underlying skeletal disease represent characteristic extra-skeletal manifestations supporting the diagnosis (9).

Radiographic features- Plain film and CT

- Intramedullary or subcortical lucent foci may be the earliest manifestation (6)
- This progresses to profound osteolysis with resorption of affected bone and lack of compensatory osteoblastic activity or periosteal reaction

Scintigraphy-$Tc^{99}m$ bone scan may initially be positive but later becomes negative with ongoing bone resorption.

It may be confused with 1) Infection2) Cancer (primary or metastatic) 3) Inflammatory or endocrine disorders.

Conclusion

In conclusion, provided that the etiopathology of vanishing bone disease has not been fully clarified, further

research is needed for more effective therapeutic interventions. Physicians should be aware of the existence of this rare entity and reliably direct affected patients to correct diagnosis and therapeutic approach.

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