

Gorham's Disease — A case report

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Introduction

Gorham's disease (disappearing bone disease, massive osteolysis, idiopathic osteolysis, essential osteolysis, progressive atrophy of bone, spontaneous absorption of bone, phantom bone, hemangiomas/lymphangiomas of bone, progressive osteolysis) is a rare disorder characterized by proliferation of vascular channels that results in destruction and resorption of osseous matrix¹. Since the initial description of the disease in 1954, fifty years have elapsed but still the precise etiology of Gorham's disease remains poorly understood. Herewith we report a case of Gorham's disease with its histopathology features.

Case report

A young girl came to our institution in 2002 at age 18 complaining of right forearm pain for one and half years. Her pain was not responding to conventional analgesics. Radiographs showed complete loss of bone of proximal one third of the ulna. In addition there were ill defined radiolucent foci of osteolysis present with in the distal humerus and proximal two third of radius involving intramedullary and subcortical regions, resembling findings seen in patchy osteoporosis. The intervening elbow joint afford no protection to extension of the disease (Fig 1a, b). A biopsy from the same area revealed few dead bony fragments along with skeletal muscle and hemorrhage. Magnetic resonance imaging (MRI) showed cystic replacement of distal humerus and proximal radius and ulna with no significant soft tissue component. T1-weighted-spin echo MRIs showed uniformly low signal intensity in the involved bones, whereas increased signal intensity was observed in T2-weighted-spin echo images. There were vanishing borders suggestive of

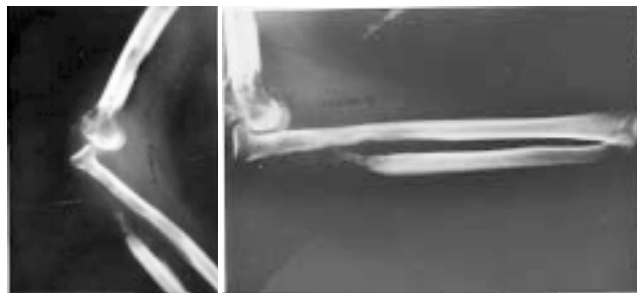


Fig 1a. An oblique view of elbow joint including distal humerus and proximal radius and ulna shows complete loss of bone of proximal one third of the ulna, along with radiolucent foci of osteolysis present with in the distal humerus and proximal two third of radius involving intramedullary and subcortical regions, b. Lateral view of elbow joint displays similar abovementioned findings.

hemangiomas lesion. Subsequently surgery was planned and above elbow amputation had been done including right distal humerus area and forearm. On gross examination, only a small fragment of humerus remained intact. Proximal part of the right radius and ulna was virtually absent. Microscopy showed numerous ectatic variable sized thin walled vascular channels surrounded by meager hematopoietic elements and adipose tissue (Fig 2a, b). At one focus there was increased osteoblastic and osteoclastic activity seen. Based on all these findings a diagnosis of Gorham's disease was given.

On follow up patient was doing well for two years after treatment.

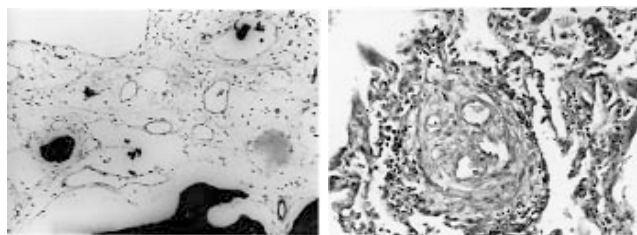


Fig 2a. Low power microphotograph shows numerous ectatic variable sized thin walled vascular channels surrounded by meager hematopoietic elements; spicules of dead bone fragments are also seen (H&E Stain 10x), b. High power microphotograph shows soft tissue hemangiomas process (H&E stain 40x)

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Discussion

The etiology of Gorham's disease is still very speculative, the prognosis unpredictable, and any effective therapy still unknown. The pathological process is the replacement of normal bone by an aggressively expanding but non-neoplastic vascular tissue^{2,3}, similar to a hemangioma or lymphangioma. Wildly proliferating neovascular tissue causes massive bone loss. 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 unknown³. The clinical presentation of Gorham's disease is variable and depends on the site of involvement. The shoulder⁴, and the pelvis⁵ are the most common sites. It often takes many months or years before the offending lesion is correctly diagnosed. Gorham's disease can involve men or women and any age group. No familial predisposition has been found⁶. A high index of clinical suspicion is needed to arrive at an early, accurate diagnosis. The medical treatment for Gorham's disease includes radiation therapy⁷, anti-osteoclastic medications, and interferon⁸. In most cases, Gorham syndrome is only diagnosed after a pathological fracture. However in the present case, patient presented with chronic pain at the site of osteolysis. According to Patel⁹ the humerus, radius and ulna are rare sites for involvement though few case reports are there in the literature.

Histopathologically, hemangiomatous or lymphangiomatous tissue can be observed². Therefore the differential diagnosis of angiomatosis is significant; the latter has, however, a multiple extraostotic origin, in contrast to Gorham syndrome. The differential diagnosis also includes other forms of osteolysis such as familiar osteolysis, macro osteolysis or essential osteolysis.

Usually, the prognosis depends on complications, such as neurological deficits or pleural effusion. The life expectation is not altered if the extremities are involved. Several therapeutic modalities have been used in the management of Gorham's disease⁹. Surgical treatment options include resection of the lesion and reconstruction¹⁰. In general, no single treatment modality has proven effective in arresting the disease. It is worth noting that the success rate after the use of a bone graft is low. Most surgeons, based on their personal experience, have observed that the bone graft undergoes dissolution.

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