

CASE REPORTS

Rapid Idiopathic Osteolysis of the Humeral Head and Clavicle

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ABSTRACT

Idiopathic osteolysis is a very rare bone condition of unknown origin. The length of the illness usually lasts for several years. The purpose of this paper is to point out the possibility of severely rapid progression of idiopathic osteolysis and to contribute to a better understanding of the natural history of Gorham-Stout disease. We report the case of a 58-year old man whose right humeral head and lateral clavicle had disappeared as a result of massive osteolysis observed only six months after onset of the symptoms. Such rapid progression has not been reported previously. His physical examination was otherwise normal. Serum interleukin-6 (IL-6) was elevated and the other laboratory tests were normal. Radiography, computed tomography scan, bone scan, and magnetic resonance imaging (MRI) evaluation revealed extensive destruction of the right humeral head and lateral clavicle involving the acromioclavicular joint. Although the histologic features could not be confirmed because the patient refused a biopsy, it was felt that the patient satisfies the other diagnostic features of Gorham-Stout disease. Follow-up examinations revealed no further bone or soft-tissue involvement. Gorham-Stout disease should be considered in the differential diagnosis of such severely rapid progression of lytic bony lesions.

Osteólisis Idiopática Rápida de la Cabeza del Húmero y la Clavícula

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RESUMEN

La osteólisis ideopática es una condición muy rara del hueso, de origen desconocido. Usualmente la enfermedad tiene una duración de varios años. El propósito de este trabajo es señalar la posibilidad de una progresión severamente rápida de la osteólisis idiopática, y contribuir a una mejor comprensión de la historia natural de la enfermedad de Gorham-Stout. Reportamos un caso de un hombre de 58 años cuya cabeza humeral derecha así como la clavícula lateral habían desaparecido como resultado de una osteólisis masiva observada sólo seis meses después de la presentación de los síntomas. Una progresión tan rápida no había sido nunca reportada previamente. Por lo demás, su examen físico fue normal. La interleukina-6 de suero (IL-6) fue alta, y las otras pruebas de laboratorio fueron normales. La radiografía, la tomografía computarizada (escáner CT), tomografía de hueso, y el examen de imágenes por resonancia magnética (MIR) revelaron una destrucción extensa de la cabeza humeral derecha y la clavícula lateral con involucración de la articulación acromioclavicular. Aunque los rasgos histológicos no pudieron ser confirmados porque el paciente rehusó una biopsia, se entendió que el paciente satisfacía suficientemente las otras características del diagnóstico de la enfermedad de Gorham-Stout.

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INTRODUCTION

Idiopathic osteolysis, also known as vanishing bone disease, disappearing bone disease, massive osteolysis, or Gorham-Stout disease, is a very rare bone condition of unknown

origin that was defined as a specific pathologic disorder by Gorham and Stout in 1954 (1). Up to 2003, over 150 cases have been reported (2). The disease is not hereditary and has no racial or sexual predilection. It has been described in patients ranging from infancy to 75 years of age. The syndrome may occur in any bone, but common sites include the long bones, the shoulder and the pelvis. Gorham and Stout (1) emphasized the following clinical features: progressive osteolysis of one or more bones in children and young adults; history of minor trauma, often associated with a pathological fracture; and vascular malformations in the affected bones or surrounding soft tissues. Patients generally present with bony deformity, corresponding muscular weakness and localized pain. Usually, laboratory test results are within normal limits except for occasional increases in the concentration of serum alkaline phosphatase and mild anaemia (3). Plain radiographs in the early stages may demonstrate features similar to osteoporosis with multiple intramedullary radiolucencies. In the later phase, considerable cortical resorption usually occurs along with massive bone loss without respect for joint boundaries (3). Complications of Gorham-Stout syndrome range from minimal disability to death. In most circumstances, disease progression is slow. Usually, progression of the disorder spontaneously arrests (2). The differential diagnosis of Gorham-Stout disease includes skeletal angiosarcomas, eosinophilic granuloma, angiosarcoma and idiopathic multicentric osteolysis. Numerous methods of treating the disease have been tried, including surgical excision with bone grafting and prosthetic replacement, radiation, and the administration of vitamin D, androgens, amino acids, adrenal extracts, calcium, fluoride, calcitonin and bisphosphonates (4). In the early stages, radiation therapy, bisphosphonates or calcitonin can arrest the disease (5).

We report the case of a 58-year old man who presented with weakness of his right arm and rapid, massive osteolysis of the right humeral head and lateral clavicle due to Gorham-Stout disease. Complete destruction was observed only six months after onset of the symptoms. The purpose of this paper is to point out the possibility of severely rapid progression of idiopathic osteolysis and to contribute to a better understanding of the natural history of Gorham-Stout disease.

Case Report

A 58-year old man attended the outpatients department with the complaints of swelling of the right shoulder for six months, weakness in his right, dominant arm and inability to lift heavy weights for the past eight weeks. He was unable to raise his right arm above his head. No history of any trauma or bone and joint disease was found in the patient or his family. He was a nonalcoholic and there was no past history of any steroid intake, diabetes mellitus, syphilis or fever. There was minimal pain during extremes of movement. He was, however, able to perform all the activities of his daily life.

On physical examination, the right shoulder was enlarged. Active right arm mobility was impaired by muscle weakness. Limited range of motion was noted on abduction, adduction, flexion, extension, and external and internal rotation of the shoulder. There was absence of any palpable soft tissue mass. Passive movements were exaggerated. There was no pain, tenderness or erythema. Movements in the elbow and wrist of the ipsilateral side were good as well as grip strength. No other joint was involved. There were no signs of any neurological deficit, psoriasis or other cutaneous lesions. His physical examination was otherwise normal. Serum interleukin-6 (IL-6) was elevated. The rest of the laboratory tests including sedimentation rate, complete blood count, serum calcium, serum phosphorus, alkaline phosphatase, liver tests, Zeil Neilson test, antinuclear factors, renal function and angiography were all normal. Rheumatoid factor and tests for tuberculosis and syphilis were negative. Thyroxine, parathormone, cortisol and protein electrophoresis were all in the normal range.

Radiographic and computed tomographic evaluation revealed extensive destruction of the right humeral head and lateral clavicle involving the acromioclavicular joint (Figs. 1, 2). No involvement of the scapula was noted. Radiographs



Fig. 1: Plain radiograph of the right shoulder. Note massive osteolysis of the proximal epimetaphysis of the humerus and destruction of the distal clavicle.

and computed tomography scans of adjacent bones were unremarkable. The bone scan revealed no uptake in the affected areas and there was no pathologic enhancement outside the affected area. On magnetic resonance images, the extent of bone destruction was confirmed, and the remaining humeral head appeared to be replaced by well-vascularized soft tissue (Fig. 3). The patient refused a biopsy.

The patient's history, clinical findings and radiographs were consistent with massive osteolysis due to Gorham-Stout disease involving the right shoulder girdle. Because of the lack of any clinical or other supporting data for other causes, the mentioned diagnosis was considered the strongest



Fig. 2: CT scan through the level of the scapular glenoid. Disappearing of the proximal humerus is evident.

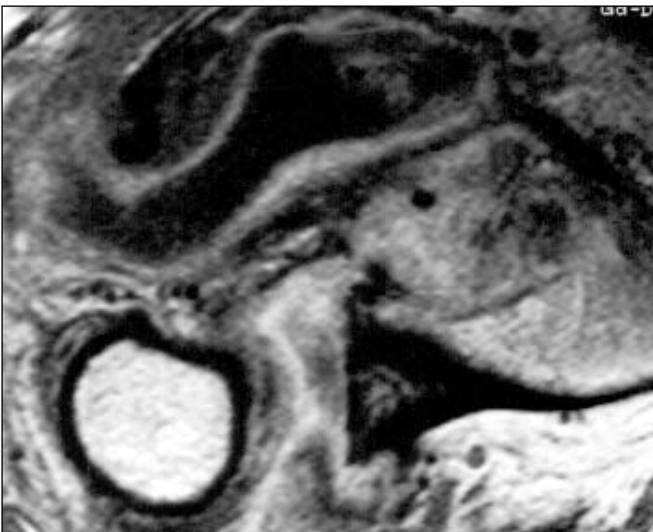


Fig. 3: Axial T1-weighted MRI of the shoulder. Significant signal increase in affected soft tissue after contrast administration. Note destruction of the humeral head.

possibility. Although the histologic features could not be confirmed, the authors felt that the patient sufficiently satisfied the other diagnostic features of Gorham-Stout disease. Follow-up examinations after 6, 12, 18 and 24 months revealed that the disease was stable with no further bone or soft-tissue involvement. We could not intervene in this patient as the destructive process had already spontaneously stopped when the patient presented to us. As there were no significant complaints about his daily activities, he was kept on regular physiotherapy. The patient is still under observation.

DISCUSSION

Idiopathic osteolysis consists of a heterogeneous group of rare diseases in which there is spontaneous onset of peripheral osteolysis with no obvious cause. MacPherson *et al* (6) classified primary osteolyses into five types: hereditary multicentric osteolysis with dominant transmission, hereditary multicentric osteolysis with recessive transmission, non-hereditary multicentric osteolysis with nephropathy, Gorham's massive osteolysis and Winchester disease (7). The lack of association with a nephropathy (8) or with a specific genetic mode of transmission differentiates Gorham-Stout syndrome from other types of idiopathic osteolytic processes (9). Gorham and Stout (10) first recognized this disease as rapidly progressive osteolysis of one or more adjacent bones resulting from excessive intra-osseous proliferation of small blood vessels or lymphatic ducts.

The aetiology and pathogenesis of this very rare disorder is still unknown. The proposed mechanisms include hyperaemic granulation tissue in the bone (11); underlying endothelial dysplasia of lymphatics, blood vessels or both (12); local hypoxia and acidosis (13); and activation of silent hamartoma (11–13). Another theory proposes that the disease is the result of deranged osteoclastic activity (5). The disease is not accompanied by any systemic symptoms, there is never a second osteolytic focus and the function of the affected limb frequently remains good for a long period of time (14). Another feature of the disease is the tendency to involve one bone only initially and then to spread frequently to adjacent bones, not respecting anatomic boundaries and readily crossing articular barriers. In more than half the cases, there was antecedent trauma (2). The localization of osteolysis is very different. It mainly affects bones that develop by intramembranous ossification (shoulder girdle, pelvis, jaw, ribs, spine), and most often is seen in children and young adults of either gender.

Clinical manifestations and complications of Gorham-Stout disease vary and depend on the affected bone and spread of pathology to adjacent tissues. Most patients describe an insidious onset of painless, bony, or overlying soft tissue deformity and instability, which progresses rapidly. The clinical course is unpredictable. In most cases, bone resorption spontaneously arrests after a variable number of years. Campbell *et al* (15) reported one case of spontaneous recovery. Despite the extensive regional loss of bone with resultant deformity and atrophy, these patients are frequently left with only mild disability. The biochemical results are usually non-specific.

Radiographically, the earliest changes are subcortical and intramedullary osteolytic areas that have no sclerotic borders (2, 16). Bone tapering at the edge of the lesion to a cone-like spicule due to progressive osteolysis, is a suggestive radiologic finding of advanced disease (17). Magnetic resonance Imaging (MRI) has only rarely been described in the diagnosis of Gorham-Stout disease. Depending on the relative amount of vascular structures and fibrosis, the

signal behaviour varies on MRI with hypointense signal on T1-weighted images and mixed, at least partially elevated, signal intensities on T2-weighted images. Soft tissue masses not necessarily seen on standard radiographs appear to be part of Gorham-Stout disease (18). The role of MRI is not to provide a specific diagnosis but rather to demonstrate the extent of disease. In accordance with previous reports, the MRI findings in the index case could be regarded as suggestive but not specific (16). Gorham-Stout disease is a diagnosis of exclusion. It is a mixture of clinical, radiological, and histopathologic findings.

Because of the rarity, the assessment of any method of treatment is difficult. At present, there is no effective therapy. Surgical interventions are frequently not possible due to pronounced osteolysis and inadequate bone for fixation. Implanted bone has been reported to be absorbed within a few weeks (19). Radiotherapy with moderate doses (40–45 Gy in 2 Gy fractions) has been reported to lead to pronounced clinical improvement (20). Other therapeutic options, including embolization, steroids, vitamin D, androgens and calcium, have not led to permanent disease control. Medical therapy with calcitonin and bisphosphonates has been proposed after the finding of highly stimulated osteoclasts in the osteolytic areas (5). Because the disease is often self-limiting, conservative treatment appears to represent an appropriate treatment option. The course of the disease is difficult to predict, but the prognosis of the condition is generally considered to be good with overall mortality of about 13% and a tendency to spontaneous arrest (19).

This case is being reported because of the extremely rapid progression of Gorham-Stout disease encompassing the humeral head and lateral clavicle and involving the acromioclavicular joint. The duration of the condition varies, but usually lasts for several years (13). The rapid progression of massive osteolysis has not been previously reported. The clinical features were suggestive of Gorham-Stout disease. On the other hand, avascular necrosis and Charcot's arthropathy can also cause a similar clinical picture.

Thus, in conclusion, idiopathic osteolysis is still an unpredictable condition. Gorham-Stout disease should be considered in the differential diagnosis of such severely rapid progression of lytic bony lesions of the shoulder. This and further new cases may well provide insight into the clinical and pathophysiological features of this disease.

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