VANISHING BONE DISEASE IN A TERTIARY TEACHING HOSPITAL IN UGANDA: A CASE REPORT

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ABSTRACT

Gorham's disease/ vanishing/ disappearing bone disease is a rare musculoskeletal disorder reported in literature. Due to its rarity, its diagnosis is usually by high index of suspicion. The management is usually individualized since there is no known standard therapy. The prognosis is generally good if diagnosed early and managed accordingly except in situations of chylothorax where there is associated increased mortality. We report a case which was diagnosed and managed successfully with a one year follow-up in a tertiary teaching hospital in Uganda.

INTRODUCTION

Gorham's disease also known as vanishing bone disease or disappearing bone disease is a rare progressive osteolysis usually affecting one or more bones in the body which can lead to partial or complete disappearance of the bone in the body (1). The pathological cause of this condition is not exactly known despite its description over 60 years ago (2).

As of 2008 about 200 cases have been reported in literature and commonly reported sites include; mandible (15%), ribs (12%), scapula (10%), humerus (8%), pelvis (10%) and femur (11%) (3). About 75% of the cases are found to be localized on a single bone and the disease process continuous progressively until it spontaneously resolves (4, 5).

Mortality associated with this condition is low except in cases of thoracic involvement (6). Due to the rare nature of this condition, a high index of suspicion will be required in order to think about or diagnose this condition especially in Africa where most hospitals do not have pathologists or radiologists. We therefore present a case of Gorham's disease which was initially thought to be a malignant disease, diagnosed in Mulago Teaching Hospital in Uganda.

CASE REPORT

A 27 year-old male presented to the Orthopaedic out-patient clinic with a 5 month history of a painful mass involving his right hip. He reported to have started experiencing pain in his right hip about 6 weeks before noticing a gradual swelling. The mass rapidly enlarged over a period of 5 months with increasing pain, aching in nature and associated with failure to walk or sit. The pain was radiating to his lower back and right knee joint. There was no history of fevers, evening chills or excessive sweating and no prior trauma to the right hip. No history of chronic illness prior to the above presentation and review of systems were unremarkable. General examination revealed a young man who appeared weak and in pain. He had moderate pallor of the conjunctiva with no jaundice, no lymphadenopathy, and no pedal oedema. On local examination, he had a diffuse mass involving the right gluteal and hip areas, warm and tender to touch and the overlying skin was tense and shiny. There was great apprehension to passive ROM of the hip joint. Distal neurovascular exam of the right leg was normal. The left hip joint was normal. Systemic examination was normal. The patient was immediately admitted to one of the orthopaedic wards in Mulago with a working diagnosis of a malignancy. A work-up plan was instituted which comprised of:

(i) Pelvic X-rays (AP view)
(ii) Chest X-ray (AP view)
(iii) Full haemogram
(iv) HIV serology
(v) Serum alkaline phosphatase
(vi) Serum calcium and phosphate assays
(vii) Open biopsy of the lesion

Figure 1
Preoperative X-ray of pelvis (AP view)

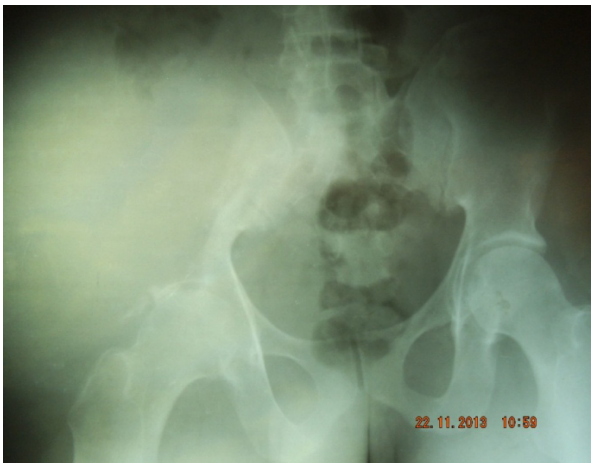


Figure 2
Postoperative X-ray (6-months after Biopsy)

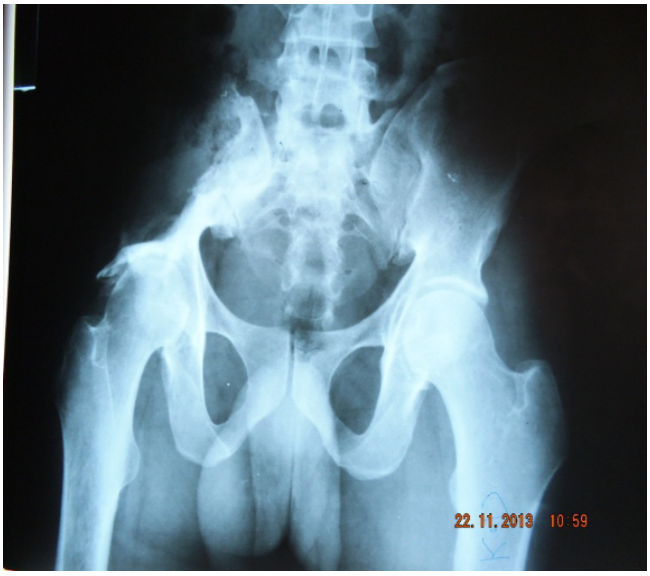


Figure 3
Preoperative chest X-ray

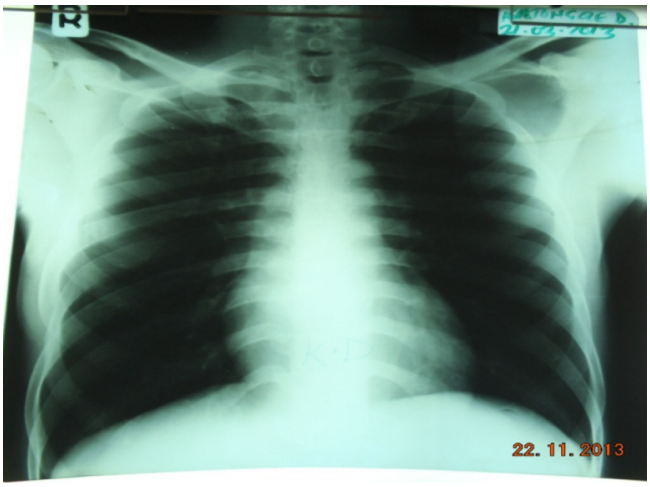
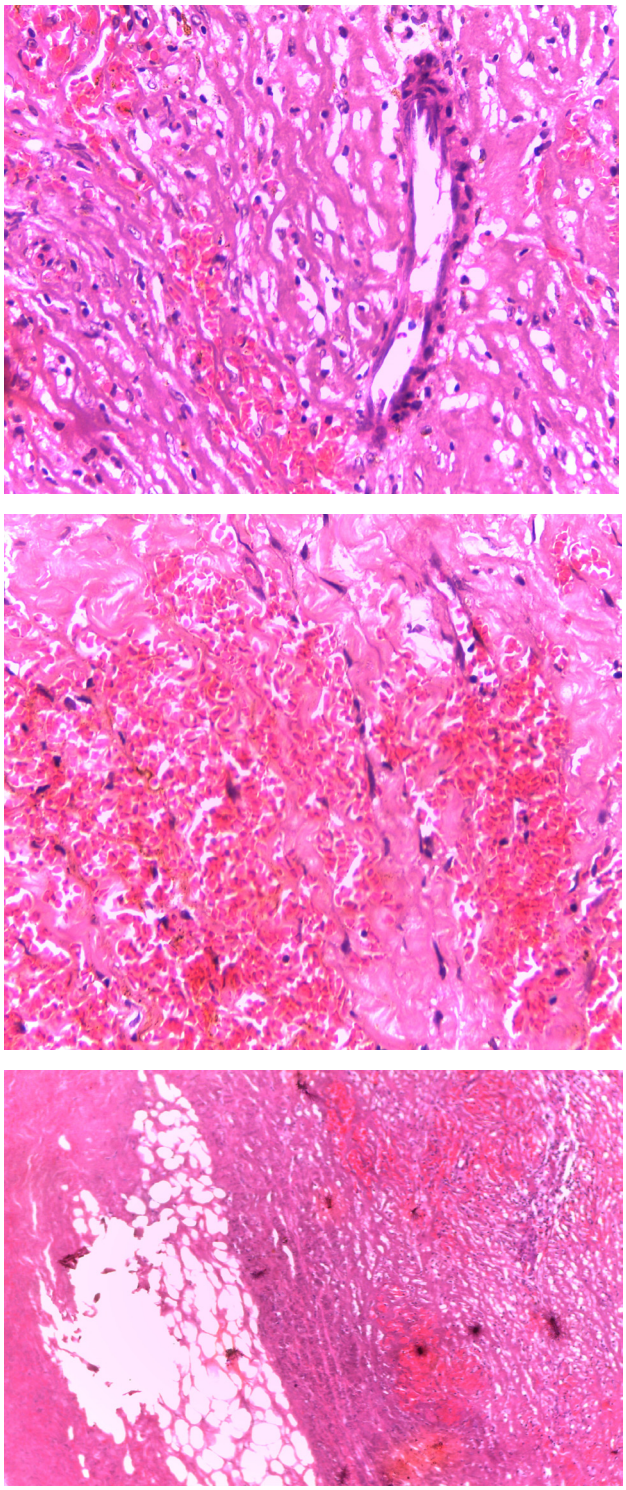


Figure 4
Patient walking 6 months post-operatively with no evidence of a mass as shown above and below

Figure 5
Showing histology slides of the biopsy done



The laboratory results revealed normal blood counts, normal serum calcium and phosphate levels and a negative HIV serology. The chest X-ray was essentially normal. However, the pelvic X-ray showed extensive lysis of the right iliac bone with sparing of the hip joint. In view of this, a working diagnosis of osteosarcoma was made and an open biopsy immediately planned. Intra-operatively, the lesion was found to have a markedly thickened capsule and on making a minor incision, there was a gush of altered blood with fibrinous debris. Samples were obtained for histology.

The patient was transfused with 2 units of whole blood and had an un-eventful post-operative recovery. He was later discharged from the ward and was put on bisphosphonate and subsequently followed-up in the out-patient clinic.

DISCUSSION

Gorham's disease is a rare musculoskeletal disorder that was first described by Gorham and colleagues in 1954 and then later Gorham and Stout discussed it comprehensively in 1955 based on their clinical experience and available case reports in literature (7, 8). Other eponyms used in literature to describe this condition include; Gorham's syndrome, Gorham-Stout syndrome or disease, massive osteolysis, disappearing bone disease, vanishing bone disease, idiopathic or progressive massive osteolysis, phantom bone disease etc (9).

The exact cause and pathogenesis of the condition is unknown. However it is said to be associated with progressive osteolysis with angiomatosis of blood vessels and lymphatics resembling that of a haemangioma or lymphangioma. It is theorized that proliferative neovascular tissue causes massive bone loss with the early stage undergoing bone resorption and replacement with hypervascular fibrous connective tissue and the late stage associated with progressive dissolution of bone leading to massive osteolysis with bone replaced by fibrous tissue (9). Also because of the hypervascularity associated with the lesion with wide capillary network of vessels it is assumed that it leads slow circulation locally leading to hypoxia and lowering of pH which favours activities of hydrolytic enzymes resulting in ossteolysis (10).

Gorham's disease can affect men and women of any age but commonly occurs in adolescents and young adults and has no gender or racial predilection (11). It can affect any anatomic site in the body but the commonly affected sites reported in literature include; mandible (15%), ribs (12%), scapula (10%), humerus (8%), pelvis (10%) and femur (11%)(3). Disease that affects the ribs, scapula or thoracic vertebra may lead to the development of chylothorax by direct invasion of the thoracic duct and without surgical intervention; it's often associated with high morbidity and mortality(9). These patients usually present with a history of an abrupt or insidious onset of pain which sometimes is triggered by a minor injury or trauma to the site, soft tissue swelling or atrophy, limitation of motion or movement, weakness of the involved limb and paraplegia in cases of vertebrae involvement with invasion of the spinal cord (9,11,12). Thoracic cage, pulmonary or pleural involvement can lead to respiratory compromise with resultant death (9). Bone infection and septic shock from Gorham's disease though rare have also been reported (13).

By way of investigations, the standard blood test are usually normal and not helpful in Gorham's disease but with slightly elevated alkaline phosphatase. Imaging modalities like plain X-rays, computer tomography scans (CT-scan), bone scans and magnetic resonance imaging have all been useful in this regard (9). Radiographically, Gorham's disease progresses through 4 stages (14). The first stage is patchy osteoporosis in the intramedullary region of bone. Second stage is development of new radiolucent areas in the periphery; third stage is associated with extra-osseous involvement evident by cortical erosion and soft tissue swelling or invasion (Figure 1). The final stage is further disappearance of adjacent bone with obvious massive osteolysis. Histologically, there is nonmalignant vascular proliferation with thin walled lymphatics and/ or capillaries surrounded by fibrous stroma between bony trabeculae and osteoclastic activity is usually not readily seen (8). Histology of this patient is shown in Figure 5. Grossly, the affected bone appears thin with an almost absent cortex enclosing a red or brown soft tissue and the bone and surrounding soft tissue may be replaced with a vascularised soft tissue mass (15). The natural course of this condition is unpredictable and an overall prevalence of mortality is 13% in literature usually due to rib involvement and chylothorax (4,16). Due to the rarity of this condition, there is no known standard treatment and it's managed on individual patient symptoms and anatomic location. Medical treatment involves, radiation therapy, anti-osteoclastic medication (bisphosphonates) and alpha-2b interferon (9). The surgical modes of treatment involve, lesion resection and reconstruction using bone graft sand/ or prostheses sometimes followed by radiation therapy (40-45 Gy in two Gy fractions) results in good clinical outcomes and few long term complications(17). In patients with chylothorax from Gorham's disease, the treatment involves, pleurodesis, thoracic duct ligation and radiation therapy etc (18).

CONCLUSION

Gorham's disease or vanishing or disappearing bone disease is a rare musculoskeletal disorder reported in literature. Due to its rarity, its diagnosis is of exclusion after thorough clinical, radiological, surgical and histological evaluation. The management is usually individualized since there is no known standard therapy. The prognosis is generally good if diagnosed early and managed accordingly except in situations of chylothorax where there is associated increased mortality.

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