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 outpatient 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 a single bone and the disease process continuous progressively until it spontaneously resolves.

Figure 1
Preoperative X-ray of pelvis (AP view)
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 osteolysis (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 development of chylothorax by direct invasion of the thoracic duct and without surgical intervention; it’s 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.
REFERENCES