GORHAM'S SYNDROME WITH PLEURAL EFFUSION AND COLONIC CARCINOMA

S E S Ng, Y T Wang

ABSTRACT

Gorham's syndrome is a rare primary disorder characterised by spontaneous bone resorption which usually arrests spontaneously. The presence of thoracic findings markedly worsens the prognosis. We report a case of Gorham's syndrome with thoracic as well as skeletal involvement who subsequently developed colonic carcinoma.

Keywords: Gorham's syndrome, pleural effusion, massive osteolysis

SINGAPORE MED J 1995; Vol 36: 102-104

INTRODUCTION

Gorham's syndrome is a rare disorder characterised by spontaneous and often progressive bone resorption[1]. Its aetiology is still not known. Biochemistry is typically unremarkable. The osteolysis usually arrests spontaneously. Prognosis is generally unpredictable. The presence of thoracic involvement is rare but is associated with a poor, if not fatal, outcome[2]. To date, there are no reported cases of carcinoma as an association. This report describes a case of Gorham's syndrome with associated pleural effusion and colonic carcinoma.

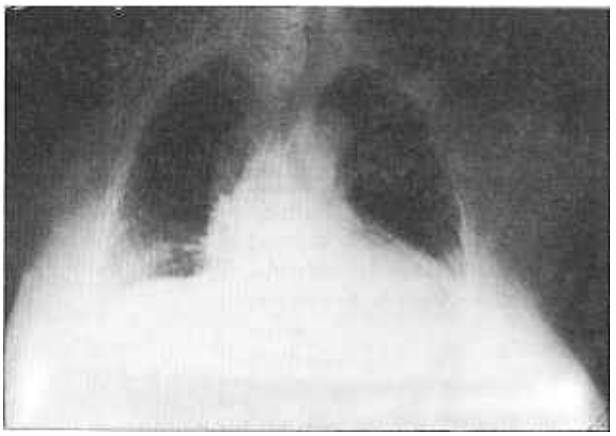
CASE REPORT

A 63-year-old woman presented to our chest unit with a four-month history of progressive breathlessness. Examination revealed a right pleural effusion. Her left upper limb was incidentally found to be swollen. It was non tender but completely flaccid. Chest X-ray (CXR) done then revealed a right subpulmonary effusion. The right clavicle and scapula could not be seen. The first and second ribs on the right were also irregular (Fig 1). X-rays of the right upper limb revealed complete absence of the humerus and ulnar. Only a faint outline of the head and terminal end of the radius was seen. There was osteopaenia of the hand bones with waisting of the metacarpal bones and phalanges. Marked soft tissue swelling was also noted (Fig 2).

The patient was subsequently admitted for a chest tap. Some 800 ml of blood stained serous fluid was aspirated. Serum as well as pleural fluid biochemistry was unremarkable. Thoracic CT confirmed the presence of pleural fluid but could not demonstrate an underlying cause. As her right arm could not be abducted, it was included in the scan, which revealed the presence of soft tissue in addition to the absence of bone (Fig 3).

Further questioning revealed that the patient was first seen eight years ago at the Orthopaedic Unit for right shoulder pain. There was no history of trauma. Over the next two years she gradually lost the use of her right upper limb. Till her present visit, she was free of chest symptoms. A review of her radiographs done in that period showed a pathological fracture of the right humeral neck with associated demineralization of the right humerus, radius and ulna. The lungs, right clavicle, scapula and hand bones were normal. A skeletal survey of the rest of the body was also normal. An open biopsy of the upper humerus was reported as foci of inflammatory cells within viable bone. Follow-up radiographs revealed increase in the demineralization of the aforementioned bones (Fig 4 and 5). Her symptoms deteriorated from pain to total inability to move the limb. She was faithful to her appointments for the first three years, following which she absconded.

Fig 1 – CXR when the patient first presented with dyspnoea.



Under the care of the chest unit, her symptoms improved after the chest tap. However she was admitted to the surgical unit for intestinal obstruction. Emergency laparotomy revealed a carcinoma of the splenic flexure. A transverse colostomy was performed, followed by a subtotal colectomy subsequently. Her progress following the colectomy was normal for the first three days. The following day, she became dyspnoeic and was found to be hypotensive. ECG showed an acute myocardial infarction and the patient died the same day.

DISCUSSION

There have been 135 cases of Gorham's syndrome reported since Jackson described the first case of massive osteolysis in 1838. This syndrome is viewed as a combined clinical, radiologic and histologic entity. Although the site of involvement is widespread, the arm and shoulder girdle is the most frequent location. The mean age of presentation is 27 years and males outnumber females by about 2 to 1[3].

The radiographic findings are quite characteristic. Bone involvement is typically monocentric and the earliest changes

Department of Radiology
Tan Tock Seng Hospital
345 Jalan Tan Tock Seng
Singapore 1130

S E S Ng, FRCR (UK)
Registrar

Y T Wang, MRCP (UK)
Consultant

Correspondence to: Dr S E S Ng

Fig 2 – X-ray of the right upper limb bones at time of presentation shows extensive resorption of the underlying bones with extensive soft tissue swelling.

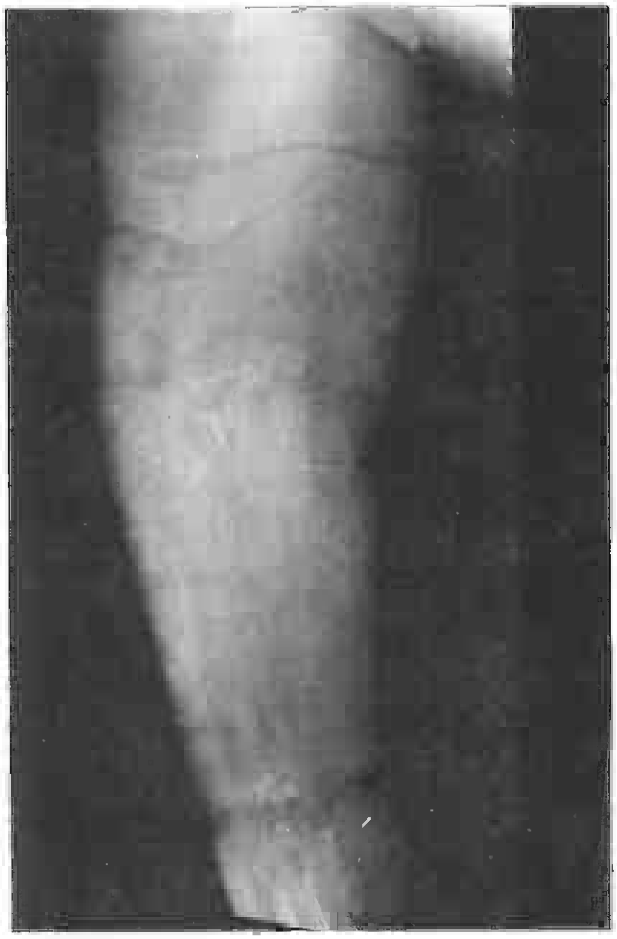
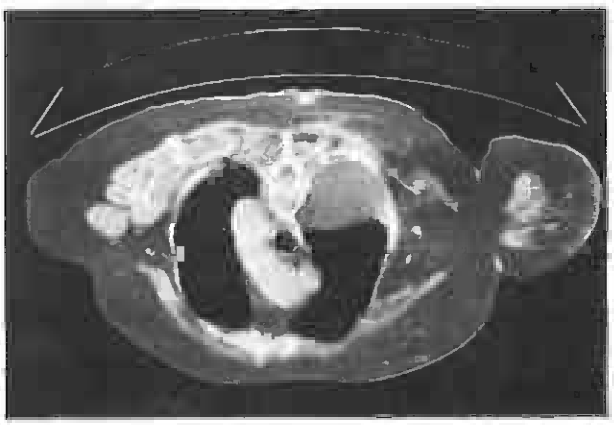
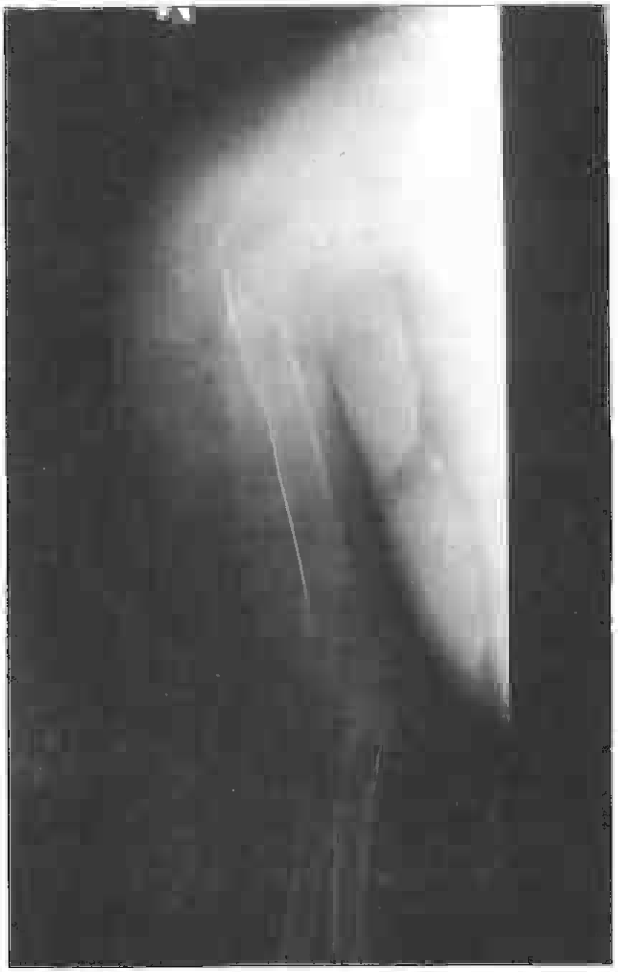


Fig 3 – Patient's thoracic CT at time of presentation.



are foci of osteoporosis. This progresses to tapering of involved long bones, resulting in a "licked candy" appearance[4]. In severe cases, complete resorption of the involved bone, as well as adjacent bones by direct spread, may be seen. The process may also extend into the adjacent soft tissues.

Fig 4 – X-ray of the right upper limb eight years prior to present admission shows fracture of the neck of the left humerus.

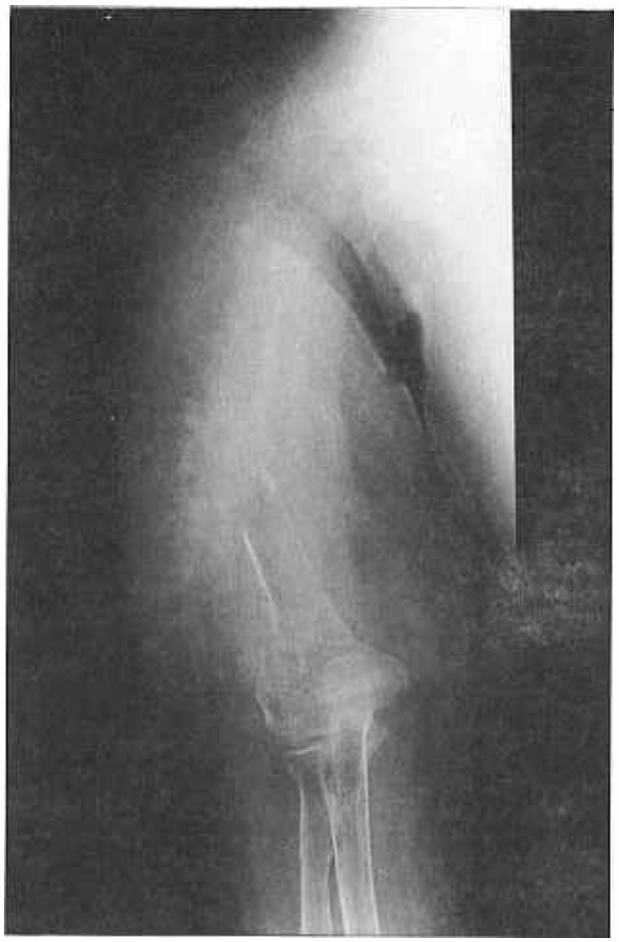


Histologically, most of the biopsies reported a benign vascular proliferation, ie bone destruction secondary to anastomosing vascular spaces lined by flattened epithelium. A less common histological finding is that of chronic inflammation[1]. This finding, which applies to our case, has led some[5] to propose that the disease is caused by pressure erosion from excessive growth of granulation tissue.

The differential diagnosis would include skeletal angioma, angiosarcoma and other forms of osteolysis. Skeletal angiomas may show some similarity histologically. Radiologically, the bony cortex as well as the surrounding soft tissue are uninvolved. The reverse applies to angiosarcoma, ie radiological features may be similar but histology will reveal malignancy. Hereditary osteolysis is associated with a strong family history as well as a young age of presentation. Secondary osteolysis arises from conditions (eg rheumatoid arthritis, syphilis and hyperparathyroidism) which run a different clinical course and have biochemical changes. Usually the osteolysis ceases spontaneously, leaving behind deformity and disability of varying degrees.

A small percentage (16%) have died as a result of the

Fig 5 – X-ray of the right upper limb four years prior to present admission showed resorption of the upper half of the left humerus.



disease[3]. The commonest cause of such deaths is chest wall involvement. Pleural effusion in particular is associated with poor prognosis[2]. There are no known reports of cases developing malignancy. On one hand the latter may be unrelated since our patient is elderly and at greater risk of developing cancer. On the other hand, the presence of chest complications may actually herald a fatal outcome or increase the likelihood of malignancy.

References

1. Gorham LW, Stout AP. Massive osteolysis (Acute spontaneous absorption of bone, phantom bone, disappearing bone): Its relation to hemangiomatosis. J Bone Joint Surg 1955; 37-A:985-1004.
2. Halliday DR, Dahlin DC, Pugh DG, Young HH. Massive osteolysis and angiomatosis. Radiology 1964; 82:637-44.
3. Nathan ND, Biscotti CV, Bauer TW, Mehta AC, Licata AA. Gorham's Syndrome: A case report and review of the literature. Am J Med 1987; 83:1151-6.
4. Torg JS, Steel HH. Sequential roentgenographic changes occurring in massive osteolysis. J Bone Joint Surg 1969; 51:1649-55.
5. Thompson JS, Schurman DJ. Massive osteolysis. Clin Orthop 1974; 103:206-11.