

# DISAPPEARING BONE DISEASE: A CASE REPORT

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## ABSTRACT

*Disappearing bone disease is a rare condition and usually affects young adults. Its aetiology is not known. A case of a 12-year-old female child, who had a fall and sustained a fracture mid shaft of femur and supracondylar region, is reported. On follow-up subsequent X-ray showed extensive osteolysis which was progressively affecting the other side of the pelvic girdle and femur. She was put on trial of calcitonin 50 IU by nasal spray for six months but there was no improvement. However, during the past three and half years the disease process had progressively extended to the other side of pelvic girdle and femur with fatal outcome. To the knowledge of the authors such progression in a short time has not been reported in any case so far.*

**Keywords:** *Disappearing/vanishing bone disease, Gorham's disease.*

SINGAPORE MED J 1994; Vol 35: 527-528

## INTRODUCTION

Disappearing bone disease is a rare condition manifested by massive osteolysis associated with proliferation of thin walled vessels within involved bones. Its aetiology is unknown and much uncertainty regarding its course, prognosis and treatment remain<sup>(1)</sup>. The disease is neither inherited nor congenital. Pain and fracture of the bones may be present and the disease seldom leads to death. Survival is worse when chest or vertebral column is affected<sup>(2)</sup>. A host of the diseases may cause osteolysis, the most common of these being infection, tumour, trauma (Sudeck's atrophy), various diseases of the nervous system, gout, the reticuloses and scleroderma. For over a century scattered case reports have described patients with "phantom bone", "cryptogenic osteolysis", "spontaneous resorption of bone", "disappearing bone", "essential osteolysis" and the like. These names are now recognised as synonyms for massive osteolysis<sup>(3)</sup>. We report a case of disappearing bone disease which started in the left femur and extended to the left pelvis and progressively involve the right pelvis and right femur in a short period with fatal outcome, which has not been reported in literature.

## CASE REPORT

A 12-year-old Malay school girl was admitted to Hospital Universiti Sains Malaysia with a history of fall from steps and following that she was unable to walk. Initially she received native treatment from a traditional healer (bomoh). This treatment consisted of massage for few weeks and

following that she developed pain and swelling in the left mid thigh. There was no other systemic abnormality. On examination, the left thigh was soft, not warm and tender. But, abnormal mobility was present. X-ray was taken of the left femur (Fig 1). There was a fracture in the mid shaft of the left femur and the supracondylar region. The cortex was thin and no new bone was seen. Blood chemistry, including parathyroid hormone, was within normal limits. The patient was treated conservatively in a Thomas splint. Follow-up

**Fig 1 – X-ray lateral view of left femur**



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X-rays showed progressive osteolysis which also extended to the left hemipelvis (Fig 2). CAT scan of the mid shaft of the left femur showed thinned out cortex without any bony expansion. Needle aspiration biopsy revealed nothing abnormal.

**Fig 2 – X-ray of both pelvis and upper part of femurs**



During follow-up it was noticed that the left lower limb appeared shorter and subsequent X-rays showed extensive osteolysis of the left femur, left hemipelvis and lower lumbar vertebrae. Other bones were normal.

Femoral angiogram was done and it was normal. Open biopsy of femur was done and submitted for histopathology examination which showed mostly bony spicules and thick fibrous bands. At places the fibrous bands were surrounded by sheets of lymphocytes. The adjacent muscle fibre bundles were diffusely infiltrated by lymphocytes. There was no increase in vascular channels. The child was mobilised initially by an above-knee caliper with pelvic band. Subsequently, the right side of the pelvis and femur was also involved (Fig 3) and she was ambulated in wheel chair. Radiotherapy at Kuala Lumpur was suggested but was refused by the parents. The patient was under follow-up for the past three years and was put on a trial of calcitonin 50 IU by nasal spray for the last 6 months. There was no improvement and the disease was rapidly progressive with fatal outcome.

**Fig 3 – X-ray of the right side of the pelvis and the femur**



## DISCUSSION

Massive osteolysis occurs predominantly in children and young adults<sup>(4)</sup>. Any bone in the body can be affected but there is a greater involvement of shoulder and pelvic girdle. The disease does not metastasize but does invade surrounding tissues<sup>(1)</sup>. Despite extensive bone resorption, normal serum calcium and alkaline phosphatase levels always indicate a lack of osteoclastic activity. The localised nature of the disease is supported by a normal erythrocyte sedimentation rate and normal blood picture. In none of the reported cases has any biochemical or endocrine abnormally been detected<sup>(4)</sup>. As the actual pathological process seen in this disorder is not yet determined, the presence of the idiopathic massive osteolysis still remains the hallmark of diagnosis. The classical histology is that of massive osteolysis - bony destruction with anastomosing vascular spaces lined by endothelium and surrounded by fibrous stroma. In the present case the predominant histology was that of fibro-collagenous bands amidst bony spicules. The features could suggest two possibilities: (i) open biopsy being done at a late stage of the disease, or (ii) the extensive osteolysis, being rapid and destructive, so that at the time of biopsy, the disease had already progressed to the fibrous stage<sup>(5)</sup>.

At present no treatment has proven effective in arresting the disease. Radiation therapy has produced inconclusive results. The possibility of spontaneous arrest precludes any conclusion regarding the efficacy of radiation therapy. It appears that the bone grafts are simply resorbed in a manner similar to that of the original bone. Braces have been used in cases of marked instability. Other types of surgical treatment have included placement of endoprostheses in cases involving long bones. Amputation and excision of isolated lesion have also been performed<sup>(1)</sup>. A new technique of using a free vascularised fibular graft was reported in a successful reconstruction of an involved femur<sup>(6)</sup>. Early irradiation of the appropriate selected volume of tissues using adequate doses of radiation can stop osteolysis and can achieve at least partial recalcification of the destroyed bone tissue<sup>(7)</sup>. Since bone resorption might be caused by osteoclastic overactivity, a trial of calcitonin by nasal spray was made to arrest further osteolysis<sup>(8)</sup>. During the past three and a half years, the disease was progressively extending to the other side of the pelvic girdle and femur. In the literature most of the patients have a prolonged insidious course with relatively normal life. However, in this case the disease was rapidly progressive with fatal outcome. Such progression has not been reported in any case so far to the knowledge of the authors.

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