

A Case of an Idiopathic Massive Osteolysis with Skip Lesions

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A patient with a 2-year history of pain in the left arm, and decreased strengths unrelieved by non-steroidal anti-inflammatory therapy, was being referred for repeating radiography. The radiologic examinations have demonstrated a unique pattern of non-contiguous osteolysis in the left elbow, proximal and distal radius, ulna, wrist, carpal bones, proximal and distal metacarpals and phalanges. Multi-site biopsies were being performed and confirmed the diagnosis of massive osteolysis. To our knowledge, this is the first case in which multifocal, non-contiguous osteolysis with skip lesions without associated nephropathy and without a hereditary pattern is being described in one extremity.

Index terms: Idiopathic osteolysis; Gorham-Stout syndrome; Disappearing bone disease

INTRODUCTION

Idiopathic osteolysis is a heterogeneous group of rare diseases in which there is a spontaneous, aggressive and extensive bone resorption. Idiopathic osteolysis was first described in 1838 and again in 1872 by Jackson (1, 2) who reported a case of "boneless arm". In 1955, Gorham and Stout (3) defined a specific disease entity and reviewed 24 cases from the literature. The Gorham-Stout syndrome presents progressive idiopathic osteolysis of one bone or contiguous bones around one focus, without respect for joint boundaries. In 1985, Hardegger et al. (4) described the most commonly accepted classification. There have

been a number of publications and case reports which do not appropriately fit into this classification (8-13).

We present a case of histologically proven massive osteolysis documented with plain radiography, CT and MRI. To our knowledge, this is the first case in which multifocal, non-contiguous, osteolytic skip lesions without associated nephropathy and without a hereditary pattern is described in one extremity.

CASE REPORT

A 38-year-old man is initially presented with pain and decreased strength in the left arm. Laboratory results were normal including renal function tests (urea, creatinine), thyroid and parathyroid hormone (PTH) tests. There was no history of any trauma and family history of similar diseases.

Radiography of upper left extremity showed no abnormalities during this time. The patient was treated with non-steroidal anti-inflammatory drugs but the pain was not relieved, the patient was referred for radiography 2 years later. During this time, radiography demonstrated almost complete non-contiguous osteolysis in the left

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Idiopathic Massive Osteolysis

elbow, proximal and distal radius, ulna, wrist, carpal bones, proximal and distal metacarpals and phalanges (Fig. 1A-D). Static images from bone scintigraphy with 99 technecium methylene diphosphate (99mTc-MDP) showed increased

activities in the osteolysed regions (Fig. 1E, F). To define the extent of the osteolysis, CT examination was performed. CT confirmed the osteolysis and accompanying soft tissue masses in the affected regions. MRI demonstrated that the



Fig. 1. Massive osteolysis with skip lesions in 38-year-old man.

Plain radiography of upper left extremity demonstrate almost complete osteolysis of left elbow (A), proximal radius (B), distal radius and ulna, wrist and carpal bones (C), proximal and distal metacarpals and phalanges (D).

soft tissue masses and the affected bones were hypointense to isointense with muscle on T1-weighted imaging (TR/TE, 732/12) and were of heterogeneous signal intensity on T2-weighted imaging (TR/TE, 3986/99) (Fig. 1G, H). The

lesions showed contrasting enhancements on T1-weighted sequences after intravenous administration of gadopentate dimeglumine.

Multi-site fine needle aspiration biopsy and tru-cut

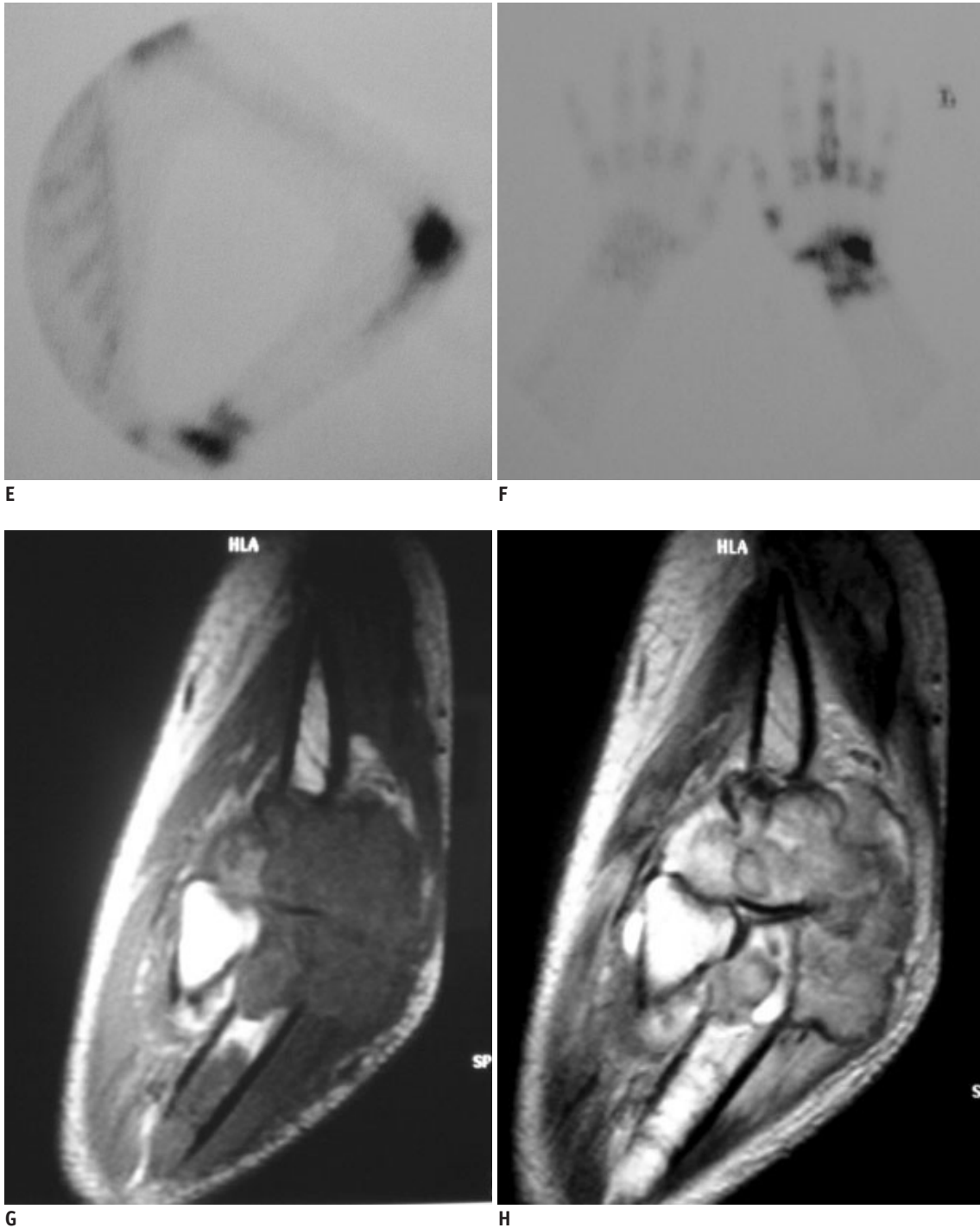


Fig. 1. Massive osteolysis with skip lesions in 38-year-old man.

Bone scintigraphy showed increased activities in osteolysed region: elbow (E), wrist and carpal bones and phalanges (F). Sagittal MR images demonstrated that soft tissue masses and affected bones were hypointense to isointense with muscle on T1-weighted imaging (G) and were of heterogenous signal intensity on T2-weighted imaging (H).

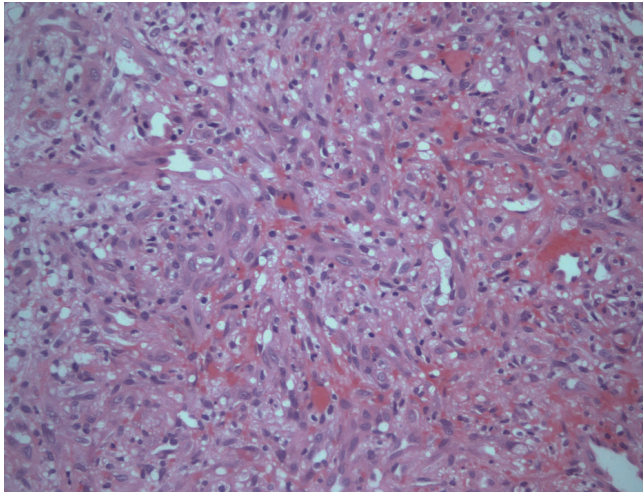
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(I) Photomicrograph (Hematoxylin and eosin, original magnification x 200) revealed fibrous tissue containing proliferated vascular structures replaced with bone trabeculae and bone marrow.

biopsy were undertaken; bone specimens were taken from proximal, distal radius and proximal ulna for pathological examinations. This revealed fibrous tissue containing proliferated vascular structures replaced with bone trabeculae and bone marrow (Fig. 1I). The pathological specimen obtained at open biopsy of a lesion in the left proximal radius also confirmed such findings. The patient was referred for radiation therapy. Radiation therapy was utilized to control the tumors locally and to alleviate associated pains. Because of its solely palliative role, full disease resolution was never accomplished. After one year follow-up there was no radiologic change.

DISCUSSION

Osteolysis, a localized resorption of bone, is a common radiological observation. Usually the destruction is associated with some underlying diseases. Gorham disease must be distinguished from osteolysis secondary to other pathologic processes, including the hereditary, metabolic, neoplastic, infectious and immunologic etiologies. Common differential diagnoses included hereditary multicentric osteolysis, essential osteolysis with nephropathy, metastasis, osteomyelitis, and rheumatoid arthritis. The clinical findings are usually helpful when ruling out these diseases (5, 6).

Idiopathic osteolysis is a heterogeneous group of rare diseases in which there is spontaneous onset of mostly

peripheral osteolysis without obvious reasons. Different classifications based on the genetic transmissions of the osteolysis have been proposed. Hardegger et al. (4) described the most commonly accepted classification with five types of idiopathic osteolysis. Type I, hereditary multicentric osteolysis with dominant transmission is commonly seen at early childhood with complaints of spontaneous pain and swelling beginning in the hands and feet. Carpotarsal osteolysis occurs over the period of a few years. Progression ceases normally during adolescence. Type II, hereditary multicentric osteolysis with recessive transmission is similar to type I except for possible associations with severe generalized osteoporosis. Type III, is a non-hereditary multicentric osteolysis with nephropathy appearing in childhood. Carpal and tarsal bones at a lesser degrees are involved. Deaths usually occur secondary to nephropathy with proteinuria and malignant hypertension. Type IV, Gorham-Stout syndrome is a monocentric syndrome which involve any part of the skeleton. Rarely, two or more anatomic regions are affected, separated by normal osseous structures. Apparently, any bone can be involved, including the small tubular bones of hands and feet, spine, skull and the mandible. (7). Unlike the other types, it may start at any age. It has neither a hereditary pattern nor an associated nephropathy. The disease is benign and the osteolysis usually stops after a few years. The Gorham-Stout syndrome presents as progressive idiopathic osteolysis of one bone or contiguous bones around one focus without respect for joint boundaries. While contiguous bony involvement is typical, multiple foci with skip lesions have not been reported in the literature (14). Type V, Winchester syndrome is a rare childhood carpotarsal osteolysis syndrome with autosomal recessive transmission. It is associated with contractures, shortness of stature, skin lesions, corneal clouding and osteoporosis without nephropathy.

This patient, without any underlying conditions, was diagnosed as idiopathic osteolysis based on radiologic, morphological and clinical findings. However, our case does not readily fit in to the classification of idiopathic osteolysis according to Hardegger et al. This case fits most closely into the type IV of Hardegger classification, however multifocal, non-contiguous osteolysis with skip lesions differ from the usual characteristics of Gorham syndrome. There have been increasing numbers of cases which cannot accurately be placed within this classification (8-13). White (8) reported four patients with multifocal osteolysis

in association with severe skin lesions. Beals and Bird (9) observed one case of carpotarsal osteolysis without associated nephropathy and a hereditary pattern. Tookman et al. (10) described a woman with osteolytic lesions of the metacarpal and metatarsal bones and terminal phalanges without a family history or renal involvement; Burkhard et al. (13) and Tauro (11) noted cases of 'multicentric Gorham-Stout syndrome' and Downing et al. (12) published one case of multicentric osteolysis in both hands and feet, especially of the phalanges, associated with normal renal functions and with no genetic basis.

We report a unique pattern of involvement in the upper left extremity with non-contiguous, multifocal osteolysis. This case is an addition to the enlarging group which cannot be readily placed in the proposed classification of idiopathic osteolysis.

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