

## Polyostotic Paget's Disease

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### ABSTRACT

Paget's disease of bone is a common metabolic bone disease in most of the European countries. The disease has distinct geographical distribution being rare in Asia. We report Paget's disease in a 70 years old Pakistani male who presented with history of pain in right leg with difficulty in walking for the last 5 months. Plain X-ray raised suspicion of Paget's disease of bone. Technetium-99m MDP bone scan showed involvement of multiple bones. Bone biopsy confirmed the diagnosis. This case illustrates that Paget's disease of bone does exist in Pakistan.

**Key words:** *Paget's disease. Bone scan. Polyostotic disease. Metabolic bone disease. Leg pain. Osteitis deformans.*

### INTRODUCTION

Paget's disease of bone (PDB) was first described by Sir James Paget under the term osteitis deformans in 1876. It is a chronic progressive disorder characterized by rapid bone resorption and deposition resulting in a mosaic pattern of lamellar bone with extensive local vascularity and fibrous tissue in marrow. This excessive breakdown and formation of bone tissue can cause bone to weaken.<sup>1</sup>

The exact aetiology of Paget's disease remains uncertain. Genetic factors play an important role in PDB, and mutations have been identified in four genes that cause Paget's disease and related syndromes.<sup>2</sup> Environmental factors also contribute to Paget's disease. Other factors which have been implicated as possible disease triggers include mechanical loading, dietary calcium and environmental toxins.<sup>3</sup>

Paget's disease of the bone appears to be more prevalent in populations of northern European origin and is thought to be rare in non-Caucasians.<sup>4</sup> In Asia, especially in India, the disease is rare.<sup>5,6</sup> In Pakistan this disease appears to be very rare but it might be because the cases here are underreported. The reported patient might not be the first person suffering from this disease, however, to the best of authors' knowledge it is the first reported case of Paget's disease of bone from Pakistan.

### CASE REPORT

A 70 years old male patient presented in April 2010 with pain in the right leg and difficulty in walking for 5 months.

There was no history of trauma, fall or fracture or local infection.

The laboratory investigations revealed elevated serum alkaline phosphatase (1122 U/L). ESR showed 50 mm fall after one hour, serum calcium was 8.23 mg/dl and PSA was 0.73 ng/ml.

Plain X-rays revealed enlarged right tibia, with bowing at its upper two third and marked thickening of cortex and trabeculae seen throughout the bone (Figure 1a).

Technetium-99m MDP bone scan (Figure 2) revealed extremely intense tracer uptake in right tibia, however, in addition right hemipelvis (Figure 1b), shaft of left fibula and proximal 1/2 of right ulna were also involved.

After whole body bone scan, X-rays of pelvis, left leg and right forearm were done which also showed similar trabecular pattern and bony enlargement of right hip bone, left fibula and right ulna. Incisional biopsy from tibial tuberosity confirmed Paget's disease of bone (Figure 3).



**Figure 1 (a,b):** a. X-rays of right leg showing enlarged right tibia, with bowing at its upper 2/3rd. Marked thickening of cortex, trabecular pattern having irregular sclerotic and lucent areas seen with two linear marginal fractures.

b. X-rays of pelvis showing enlarged right hip bone, showing patchy sclerosis and coarse trabecular pattern, loss of ileopectineal line on right side, sclerosis of right acetabular rim, and reduced joint space at the right hip joint.

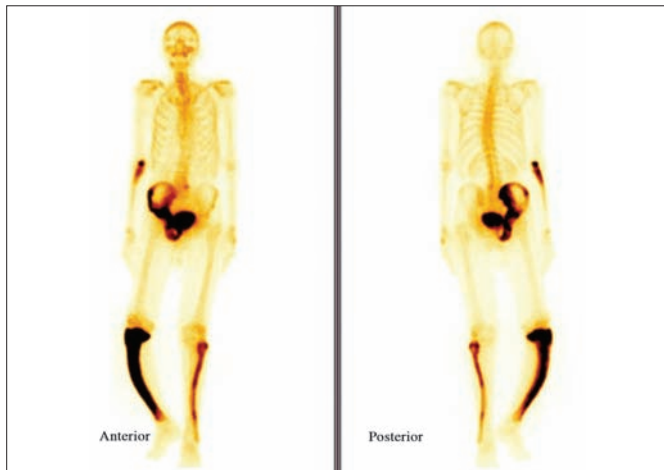
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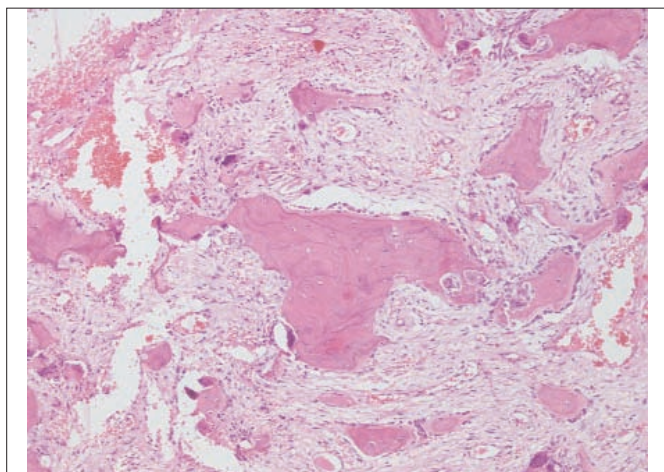
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**Figure 2:** Technetium-99m MDP bone scan revealed extremely intense tracer uptake in right tibia along with bowing of its shaft. Right hemipelvis, left fibula and proximal 1/2 of right ulna were also involved.



**Figure 3:** Photomicrograph showing trabeculae of lamellar bone with mosaic pattern in bony trabeculae, the intervening stroma is composed of vascularized connective tissue.

Oral bisphosphonates i.e. tablet alendronate 40 mg/day was started. Follow-up after 18 months showed significant improvement in pain. Serum alkaline phosphatase reduced to 795 U/L.

### DISCUSSION

The distribution of Paget's disease throughout the world shows marked racial and geographic variations and can vary in populations through migration or loss of geographic isolation. Paget's disease is relatively common in western countries; its prevalence is low in Scandinavian region. It is distinctly rare in Asia.<sup>9</sup> There are isolated case reports of PDB in Indians. Researchers have postulated that prevalence in India is underestimated as most cases were incidental findings.<sup>5,6</sup> Underreporting of cases was also one of the possible cause of low prevalence in India.<sup>7</sup>

The disease might be sporadic or familial, while looking at the comparison between Chinese and Caucasian

data; in all of the Chinese patients and in Indians the disease was sporadic.<sup>7</sup> Similar is the case here with no family history of Paget's disease, however, in Western countries 14% of patients have familial disease.<sup>1</sup>

Irving *et al.* discussed PDB in eight non-Caucasians immigrants, proposed that they might have acquired the predisposing environmental factors, if any, for the disease.<sup>4</sup>

Patient with Paget's disease of bone may present with headache and increased head size, bone pain and deformity, fracture, arthritis, spinal stenosis, cranial nerve involvement and difficulty in chewing. Rarely they may present with obstructive hydrocephalus, recurrent paraparesis and optic atrophy. Raised alkaline phosphatase in the presence of classical radiology is diagnostic of PDB.<sup>7</sup> If there is uncertainty concerning the diagnosis, a bone biopsy is the definitive means of establishing the diagnosis. Bone scan is helpful for whole body survey and for detection of asymptomatic lesions located in "at risk" areas. In a small percentage of patients, the bone scan may be positive before the appearance of an X-ray abnormality. In patients with quite localized disease and normal biochemical indices, serial quantitative bone scans may be used to determine the objective response to therapy.<sup>8</sup> Bisphosphonates therapy is highly effective in reducing symptoms and it has been shown to heal radiological lesions and restore normal histology.<sup>2</sup>

Paget's disease occurs predominantly in the elderly and its incidence increases with increasing age. The case we reported was 70 years old, which is the common age group suffering from this disease. In Caucasians, the prevalence approaches 10% by 90 years and affected individuals are rarely discovered before 20 years.<sup>9</sup>

Haddaway *et al.* found that younger patients and females had more chances of monostotic disease, and females had fewer polyostotic sites than males.<sup>10</sup> While Bhadada *et al.* found more males suffering from the disease as compared to females and more polyostotic as compared to monostotic patients.<sup>7</sup> This patient was a case of polyostotic Paget's disease involving tibia, pelvic bone, fibula and ulna. Common sites of involvement in Chinese patients were spine, sacrum, skull, pelvis, tibia and jaw.<sup>1</sup> In Indians most commonly involved bones were lumbar spine and skull followed by pelvis,<sup>8</sup> while in Western population pelvis, sacrum and spine are the most commonly involved sites, in contrast in our patient skull bone was not involved.<sup>1</sup>

In order to comment on the age, gender predilection, sites of involvement, and possible aetiology in our country we need few more cases to be reported, This case revealed that Paget's disease of bone does exist in Pakistan.

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