

Clinical Image

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Pathological fracture

Clinical Presentation

A 42-year-old Saudi lady not known to have any medical illness presented to the emergency room complaining of severe pain in the left hip after a minor trauma. She gave a history of left hip and upper thigh pain mild, dull, aching pain, especially on prolong standing, walking, and climbing stairs. Clinical examination revealed ecchymosis in the left hip and upper thigh with limitation of movement of the left leg due to pain. Laboratory investigation revealed normal full blood count, erythrocyte sedimentation rate, C-reactive protein, calcium, phosphate, urate, urea, and electrolytes and liver function tests but alkaline phosphatase (ALP) was elevated at 250 u/L (normal value: 35-104 u/L), and a slightly elevated parathyroid hormone (85) (normal value: 15-65). Plain x-ray was carried out (**Figure 1**). The fracture was fixed surgically and bone biopsy showed thickened with slightly disorganized lamellar bone with osteoclastic resorption. Bone marrow was partially replaced by fibrovascular stroma. No tumor seen and changes are consistent with those of Paget disease.

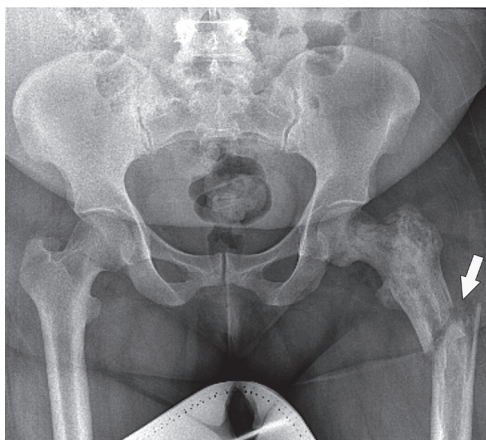


Figure 1 - An image showing the pathological fracture of the upper left femur.

Questions

1. Describe the abnormalities in the x-ray
2. What is the differential diagnosis?

Clinical Image

Answers

1. The left leg revealed thick trabeculation with sclerotic changes and expansion of the bone at the proximal left femoral shaft and femoral neck, associated with distal subcortical osteolysis and pathological fracture. Findings are suggestive of Paget's disease with pathological features, however, the underlying malignant bony lesion should be considered.
2. Paget's disease of bone (diagnosis) with pathological fractures; bone tumor is a differential diagnosis.

Discussion

Paget's disease of the bone (PD) is an uncommon chronic bone disease affecting 1% of adults in the USA. It is more common in Caucasians of Northern Europe ancestry, and rare in the Far East, African, and Middle East. It is very rare in the Saudi population.¹

It is a localized bone remodeling starting with excessive osteoclastic over activity (bone resorption) followed by compensatory osteoclastic overactivity (bone formation), this process leads to a disorganized mosaic pattern of woven and lamellar bone. Often associated with increased vascularity, marrow fibrosis, and mechanical weakness it is more susceptible to fractures. It was first described as a chronic inflammation of the bone (osteitis deformans).² It is not known what causes PD, but environmental and genetic factors have been implicated. A positive family history was reported in 12-30% of patients.³ Studies of potential genetic markers for PD found an association between human leukocyte antigen (HLA) and PD, HLA DQW1, and HLA DR2. Cytokine expression seen in PD, interleukin (IL-6) was found in the bone marrow, and peripheral blood of PD patients, this could be secondary to viral infection that upregulates IL-6.⁴ Viral infection was proposed as a possible etiology of PD by causing activation of osteoclasts, mini viruses where it is anticipated to cause this pathology after several years of infection, such as para myxovirus (measles, or canine distemper viruses). Viral inclusion particles have been identified in PD osteoclast in some studies.⁵

Paget's disease of the bone is manifested by bone pain, which is the most common presentation followed by bone deformities, such as tibial bone, and skull thickening may occur in advanced cases. The PD can affect every bone but the most commonly affected are the axial bone, lumbar spine, pelvis and femur, and also the thoracic spine, sacrum, skull, tibia, and humerus. The hands and feet are rarely affected. The PD may have other complications depending on the side affected, and this includes deafness, nerve entrapment, dental malocclusion, spine stenosis, headache, stroke, high output cardiac failure, and malignancy (osteogenic sarcoma, fibro sarcoma) and benign giant cell tumor. It can also cause hypercalcemia due to immobilization. The diagnosis of PD is usually made by symptoms and high ALP; sometimes bone-specific ALP (BSAP) can be useful. Elevated urinary markers like hydroxyproline, deoxy pyridinoline, c-telopeptide and N-telopeptide may help to diagnose patients with PD. Serum calcium and phosphate are usually normal, secondary hyperparathyroidism is seen in 10-15% of cases due to turnover was found in men with severe PD.³ Radiographic appearance in the plain x-ray will demonstrate both osteolysis, which occurs usually subchondrally in the tubular bone, and excessive bone formation. In the late stage of the disease, the lyses will diminish, and there will be mainly thick sclerotic bone. Treatment should include calcium, vitamin D supplement, and bisphosphonate. Salmon calcitonin has also been used in the treatment of PD.³

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