ADULT HYPOPHOSPHATEMIC OSTEOMALACIA

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Key-word: Osteomalacia

Background: A 32-year-old lady of Asian descent presented with pain and severe stiffness of the back and hips progressively worsening over several years. She was diabetic but had no past medical history of rickets or renal disease. On examination she was of short stature with marked thoracic kyphosis and flattening of the lumbar spine. Spinal movements were globally restricted and the hips demonstrated a fixed flexion deformity. Her initial diagnosis had been ankylosing spondylitis until she had a MRI scan. Plain radiographs and further MRI of the hips were also performed. Subsequent laboratory tests revealed she was vitamin D deficient, normocalcemic with raised parathyroid hormone (PTH) and alkaline phosphatase.

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Work-up

On radiograph of the lumbar spine (Fig. 1), anteroposterior (A) and lateral (B) projections there is loss of the normal lumbar lordosis with symmetrical marginal syndesmophytes seen anteriorly and posteriorly along the vertebra. On the lateral radiograph, sclerosis and evidence of fusion of the facet joints is noted. Bony bridging is seen in the superior aspect of the SI joints.

On radiograph of the cervical spine, lateral projection (Fig. 2), there are prominent anterior non-bridging osteophytes noted in the cervical vertebra with minimal narrowing of the intervertebral disc space.

On MRI of the lumbar spine, sagittal T1-weighted sequence (Fig. 3), anterior and posterior marginal syndesmophytes is seen in the lumbar spine. High signal ‘shiny corners’ reminiscent of Romanov’s sign is also noted mimicking ankylosing spondylitis.

On radiograph of the pelvis, frontal projection (Fig. 4); bony proliferative change is seen on the superolateral aspect of the acetabulum bilaterally. There is also ‘whiskering’ of the ischial tuberosities. Bilateral over-coverage of the femoral heads is also noted in the hip joints.

Radiological diagnosis

The radiological appearance is of adult hypophosphatemic osteomalacia.

Discussion

Osteomalacia is a disorder of insufficient osteoid mineralization causing bone softening in mature cortical and cancellous bone. This is largely due to dietary deficiency, decreased absorption or metabolic pathway disruption of vitamin D.

Vitamin D deficiency is predominantly due to inadequate sunlight exposure coupled with a diet deficient in vitamin D. The impact of vitamin D deficiency on the body’s homeostasis is to cause a secondary hyperparathyroidism with resultant phosphaturia and hypophosphatemia. Hypophosphatemic osteomalacia is one of the rarer causes of osteomalacia. The most common cause of hypophosphatemic osteomalacia is an inherited X linked abnormality (also known as Vitamin D-resistant osteomalacia), in which the renal tubules inefficiently reabsorb phosphate. Phosphate wasting is the basis of the affected individual’s inability to establish normal ossification. Genetic mutation of the PHEx gene of osteocytes and mature osteoblasts, which leads to this disorder, can be sporadic in up to a third of cases. Hypophosphatemic osteomalacia can be oncogenic or tumor associated, with mesenchymal hemangiopericytoma of bone or soft tissue being the main culprit. Fanconi’s syndrome and renal tubular acidosis can also cause this disorder. Our case showed no evidence of these rare etiologies.

The clinical features of adult hypophosphatemic osteomalacia are variable with patients presenting with short stature, lower limb deformities and limitations of movement. Vague musculoskeletal pain and tenderness as well as pain due to complications such as insufficiency fracture can be present. Proximal muscle myopathy associated may be present, as in our case.

Laboratory findings typically include normocalcemia, hypophosphatemia and normal vitamin D levels. Our case differed only in the low vitamin D levels which responded to supplementation but with no clinical improvement.

Imaging manifestations include normal or increased bone mineralization, extensive enthesopathy (bone proliferation at sites of ligament, tendons, joint capsule and interosseous membrane) and occasionally insufficiency fractures.

Syndesmophytes, squaring of vertebral bodies, spinal facet and sacroiliac joint fusion may occur which can cause the radiographic appearance to resemble that of ankylosing spondylitis as occurred in this case. Generalized enthesopathy raises the differential diagnosis of diffuse idiopathic skeletal hyperostosis (DISH) which can be difficult to differentiate from hypophosphatemic osteomalacia with which it can indeed co-exist. However, the absence of diffuse anterior longitudinal ligament hyperostosis in this case makes it less likely.

In our case, MRI of the spine hips and careful review of associated radiographs did not display any insufficiency fractures but was helpful in displaying the widespread enthesopathy of the lumbar spine and pelvis typical of adult hypophosphatemic osteomalacia. The patient’s management was altered with vitamin D and calcium supplementation achieving subsequent biochemical improvement but not full symptomatic relief.

Bibliography