Disabling Enthesopathy Sequelae of Hypophosphatemic Rickets/Osteomalacia

Hipofosfotemik Rikets/Osteomalazinin Sakat Edici Entesopati Sekeli

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ABSTRACT

The patient with X-linked hypophosphatemia typically presents with short stature, with possible bone deformities. In patients with X-linked hypophosphatemia, diffuse bone sclerosis and severe generalized enthesopaties may be present as radiographic manifestations. These manifestations may severely restrict the functional range of motions. With this case we wanted to draw attention of clinicians that X-linked hypophosphatemia may present with severe complications which should be well differentiated from other clinical scenarios in elderly. (*FTR Bil Der 2011;14: 96-8)*

Keywords: X-linked hypophosphatemic rickets, enthesopathy, radiographic findings

ÖZET

X'e bağlı hipofosfatemili hastalar genellikle kısa boylu, kemik deformiteleri olan hastalardır. X'e bağlı hipofosfatemide yaygın ve belirgin osteoskleroz ve entesopatiler görülebilir ve hareket kabiliyetini ileri derecede kısıtlayabilir. Biz, X'e bağlı hipofosfatemili yaşlı hastaların diğer klinik durumlardan ayırtedilmesini gerektiren ciddi komplikasyonlarla gelebileceği konusunda klinisyenlerin dikkatini çekmek istedik. (*J PMR Sci 2011;14: 96-8)*

Anahtar kelimeler: X'e bağlı hipofosfatemik rikets, entesopati, radyografik bulgular

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Introduction

The patient with X-linked hypophosphatemia (XLH) typically presents with short stature, with possible bone deformities. Pain in these patients may be due to an insufficiency fracture or enthesopaties. In patients with XLH, diffuse bone sclerosis and severe generalized enthesopaties may be present as radiographic manifestations (1, 4, 5). These findings are more pronounced in elder patients (1). Spinal facet and sacroiliac joint fusion may occur, which may cause the radiographic appearance that resembles ankylosing spondylitis (AS) (3). Diffuse involvement of the anterior longitudinal ligament of the thoracic and lumbar spine is rare in patients with XLH (2). These manifestations are of considerable clinical importance in that they may limit functional range of motion and are often painful. The characteristic findings of osteosclerosis and diffuse pronounced enthesopathy in adults indicate X-linked hypophosphatemia.

We describe a 65 years old man who had short stature with lower extremity bone deformities and severe limitations

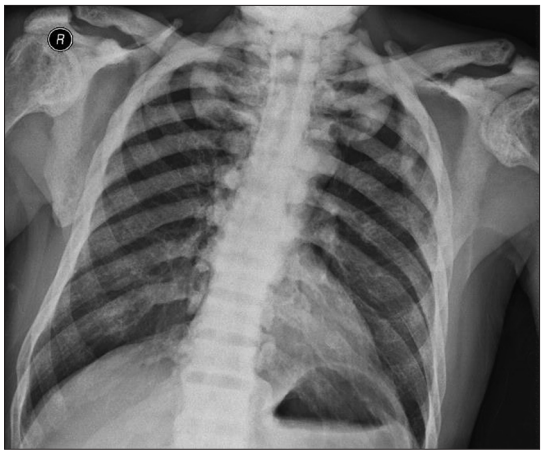


Figure 1. The patient of short stature with lower extremity bone deformities

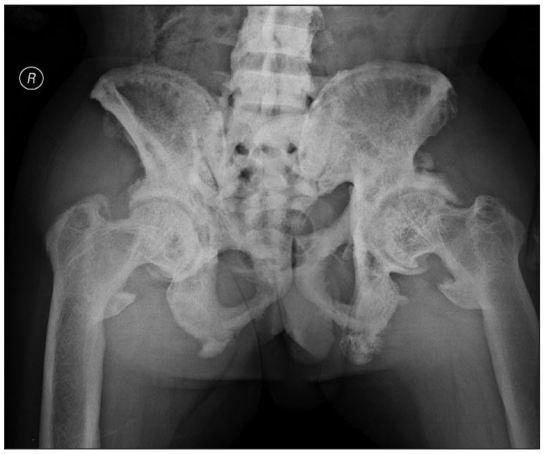


Figure 2. Bone scan from the patient demonstrating several areas of increased uptake, including the shoulders, elbows, wrists, right sternoclavicular joint, T6, T7, T8, T9, T11, T12, femoral heads, knees, right ankle. Also note the presence of lower extremity deformities

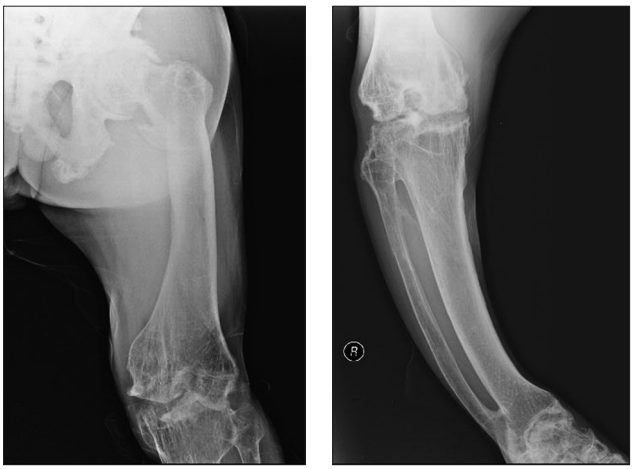


Figure 3-4. Radiographs of the upper (fig.3) and lower (fig.4) leg showed bowing of the femur and tibia/fibulae, respectively, and diffuse osteopenia of long bones. Also note the radiographic features of rickets including metaphyseal widening and cupping of distal femur and tibia/fibulae (fig 3 ve 4)

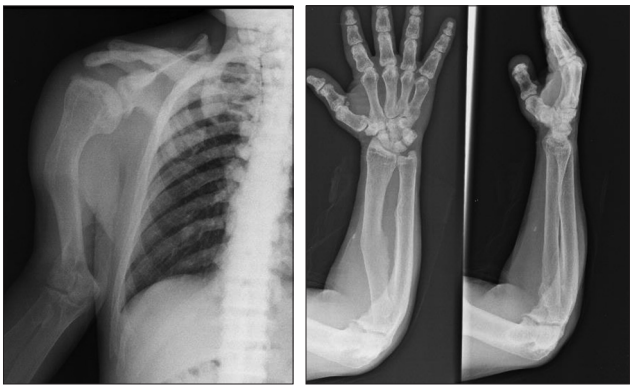


Figure 5-6. Radiographs of the upper (fig.5) and lower (fig.6) arm showed bowing of the humerus and radius/ulna, respectively, and diffuse osteopenia of long bones

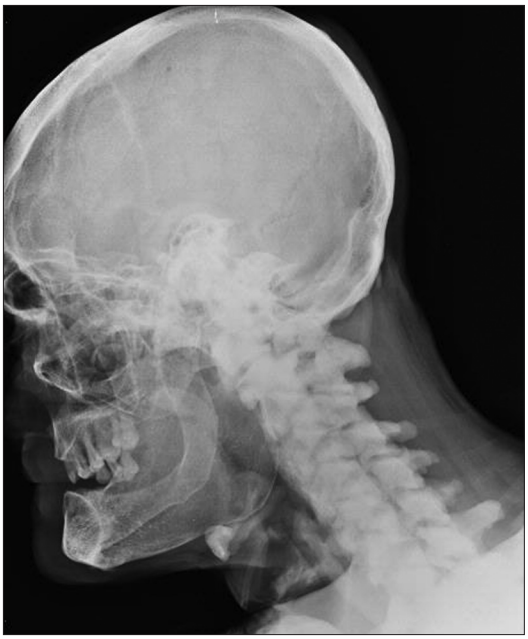


Figure 7. Cervical vertebral and temporal osteosclerosis

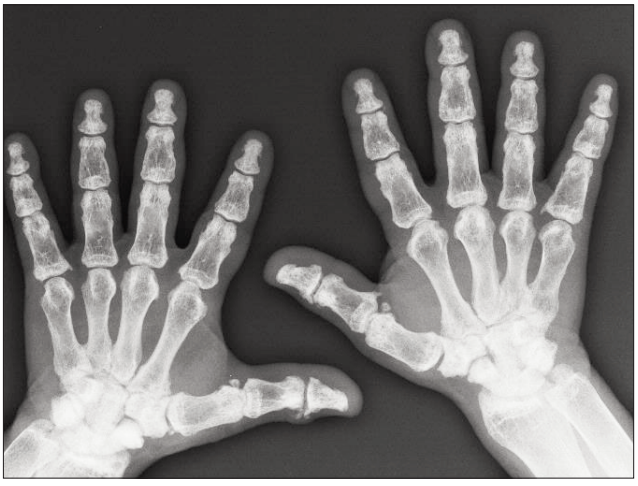
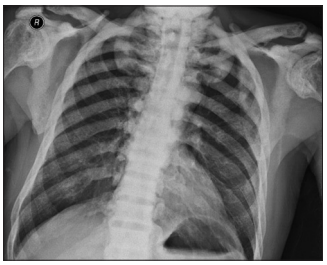


Figure 8. Carpal osteosclerosis



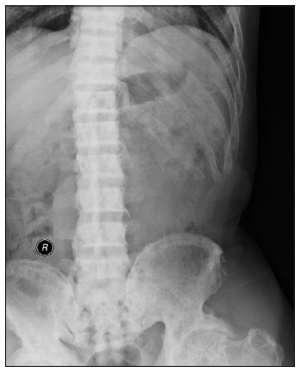


Figure 9-10. Anteroposterior chest (fig.9) and lumbar spine (fig.10) radiographs shows pronounced enthesopathy and osteosclerosis

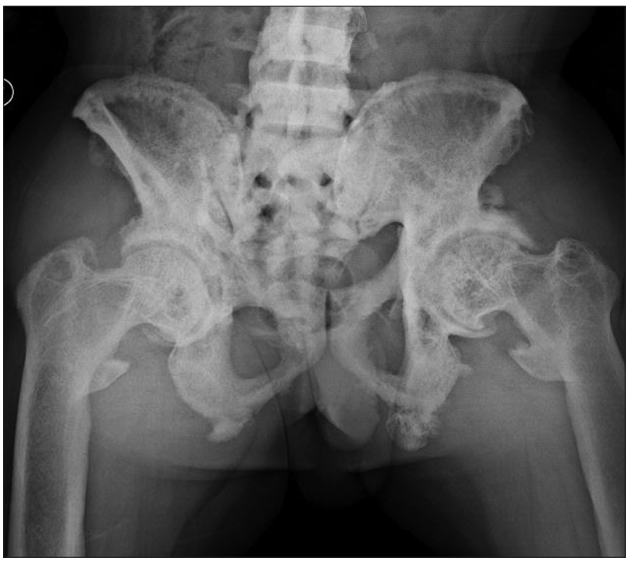


Figure 11. Anteroposterior pelvic and hips radiographs demonstrate pronounced enthesopathy. Note sacroiliac joint sclerosis represents ligamentous calcification and ossification

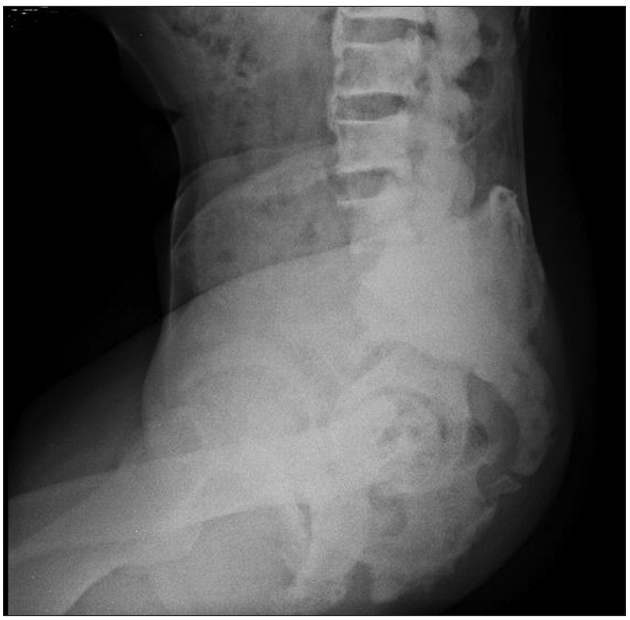


Figure 12. Lateral lumbar spine image shows enthesopathy of the anterior vertebral bodies and osteosclerosis

of movement. He had been told that the lower extremity deformity was congenital. He had no family history of bone disease.

In physical examination, his neck movements in all directions were severely limited and his neck had ankylosis at flexion posture. His low back, hip and knee movements in all directions were moderately limited. Ankle and subtalar joint movements in both extremities were severely limited. He could able to walk only short distances with difficulty.

Laboratory analysis revealed; serum creatinin 0.7 mg/dL (N, 0.5-1.25), calcium 9.1 mg/dL (N, 8,4-10,2), phosphorus 1,4 mg/dL (N, 2.3-4.7), alkaline phosphatase 167 lU/L (N, 40-150), parathormone 87.7 ng/dL (N, 15-68.3), 1.25-dihydroxyvitaminD 29.99 pg/mL (N, 19.6-65), 25-hydroxyvitamin D3 level 28.2 ng/dL (N, 15.7-60.3). Urinary calcium/creatinine excretion ratio (0.09 mg/mg) was normal. The tubular maximum reabsorption of phosphate per glomerular filtration rate (5,8) was decreased.

In imaging he had no insufficiency-type stress fracture. Radiographs of the upper and lower extremity showed bowing and diffuse osteopenia of long bones. Spine (fig.1) and pelvic (figure 2) radiographs shows pronounced enthesopathy and osteosclerosis.

Although no genetic evidence, we considered that the patient was XLH due to the presence of clinical, biochemical and radiological findings. He was started on treatment with oral phosphate 500mg three times daily and 0.25 µg calcitriol twice daily, which was subsequently increased to 0.5 µg twice daily.

As in our case, in elderly the XLH may present with very typical and pronounced osteosclerosis and enthesopathies. The latter may also seen in AS which should be considered in differential diagnosis. With this case we wanted to draw attention of clinicians that XLH may present with severe complications which should be well differentiated from other clinical scenarios in elderly.

References

1. Pitt MJ. Rickets and osteomalacia. In:Resnick D, ed. Diagnosis of bone and joint disorders. 4th ed. Philadelphia, Pa: Saunders, 2002; 1901–1945.
2. Polisson RP, Martinez S, Khoury M, et al. Calcification of entheses associated with X- linked hypophosphatemic osteomalacia. N Engl J Med 1985;313:1-6.
3. Burnstein MI, Lawson JP, Kottamasu SR, Ellis BI, Micho J. The enthesopathic changes of hypophosphatemic osteomalacia in adults: radiologic findings. AJR Am J Roentgenol 1989;153:785-90.
4. Liang G, Katz LD, Insogna KL, Carpenter TO, Macica CM. Survey of the enthesopathy of X-linked hypophosphatemia and its characterization in Hyp mice. Calcif Tissue Int 2009;85:235-46.
5. Reid IR, Hardy DC, Murphy WA, Teitelbaum SL, Bergfeld MA, Whyte MP. X-linked hypophosphatemia: a clinical, biochemical, and histopathologic assessment of morbidity in adults. Medicine (Baltimore) 1989;68:336-52.