


**Inflammatory polyenthesopathy in a patient with X-linked osteomalacia**

Sirs,

X-linked hypophosphatemia (XLH) due to a renal tubular defect in phosphate transport and a mutation of the phosphate regulating gene, is the most common inherited form of rickets in developed countries (1-3). While the main radiographic feature in childhood is rickets, in adult life one predominantly sees generalised calcific enthesopathy (1, 4-6). Polisson et al. (1) suggested that the enthesopathy of XLH cannot be attributed to the other two common causes of generalised enthesopathy, i.e. diffuse idiopathic skeletal hyperostosis (DISH) or spondyloarthritides, and, on the basis of histologic evaluation performed in selected patients, excluded the possibility of an inflammatory form. However, we would like to report the case of a patient with XLH who presented many aspects suggesting an inflammatory enthesopathy.

A 42-year-old male patient presented with a 5-month history of diffuse joint pain and stiffness, mainly involving the shoulders and knees, which started abruptly after a long bicycle ride. The patient had been affected since the age of 18 months by an XLH for which, from the age of 5, he underwent several osteotomies in the legs. Treatment with phosphate and low dose vitamin D was introduced at the age of 15 and continued for 5 years. He was then well until the present episode. The family history was negative.

Physical examination revealed short stature (163.2 cm), bow legs, and loss of range of motion in the hips, knee, and shoulders without significant swelling, but with tenderness and warmth over the shoulders and knees. Laboratory investigations revealed only a slight elevation of the ESR (35 mm/h) and a CRP of 1.7 mg/dl (normal < 0.6 mg/dl). X-rays detected extensive calcified enthesopathies on the supero-lateral patella, femoral metaphysis, and pelvis, and multiple areas of hyperostosis in the tibia and fibula. In the left femur the diaphyseal bowing was associated with a mid-diaphyseal lateral active Looser-Milkman zone. Bone scintigraphy (Fig. 1) revealed intense uptake mainly in the left knee, right coxo-femoral area, and a small area in the diaphysis corresponding to the Looser-Milkman zone. X-rays clearly demonstrated features of an enthesopathy that, due to the abrupt onset of symptoms, the elevated ESR and CRP, and the intense activity of the clinically affected areas shown on bone scintigraphy, seemed to be of the inflammatory type. However, no sacroiliac involvement or HLA-B27 were found. The patient was treated with 150 mg/day of sodium diclofenac for 3 months, and experienced a satisfactory recovery from his symptoms. Two years later he experienced some severe flares, mainly in the shoulder and knees, which occurred every 2-3 months and lasted approximately one week. In 1985 Polisson et al. (1) reported that 69% of 26 patients with XLH were affected by generalised enthesopathy. These aspects should be considered as an integral part of XLH and are very frequent, being found in 33% of patients under the age of 30 with XLH, and in all those over this age (7).

The pathogenesis of the enthesopathy associated with XLH is unclear and seems to be unrelated to therapy. Unlike DISH, it is relatively symmetric and does not show significant spinal ligament hyperostosis. The differential diagnosis from spondyloarthritides was made based on the absence of sacroiliac involvement, subchondral bone erosions or syndesmophytes. Thus, the inflammatory features found in our patient are unusual and difficult to explain. A possible cause could be the disruption in tissues surrounding the microcrystals contained in calcified deposits of the enthesopathies, probably induced by traumatic or metabolic changes, in our patient provoked by an unusually long bicycle ride.

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References


