X-linked Hypophosphatemia (XLH) Mimicking Rheumatic Disease

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A 38-year-old woman, who had been genetically diagnosed with X-linked hypophosphatemia (XLH) in childhood, was referred to our hospital due to persistent back pain and bilateral heel pain. She had visited an orthopedic surgeon and had been treated with adalimumab under a diagnosis of ankylosing spondylitis. A detailed interview revealed that she had ceased taking phosphorus and vitamin D. An X-ray showed enthesopathy, ligament calcification (A), and vertebral

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spinal osteophytes (B), and Looser’s fracture (C). Her serum fibroblast growth factor (FGF)-23 level was increased (68 pg/ml).

XLH is a type of hereditary hypophosphatemic rickets/osteomalacia caused by defective proximal tubular reabsorption of phosphate, which involves the PHEX gene (1). The complications in adults are often difficult to distinguish from rheumatic and spinal diseases (2). Continuation of treatment with phosphorus and vitamin D, and treatment with monoclonal antibodies against FGF-23 can be effective for ameliorating orthopedic symptoms (1). A past history of orthopedic surgery, bone changes and increased FGF-23 are diagnostic for XLH.

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References


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