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CASE REPORT

Hypophosphatemic Rickets with Sacroiliitis-like Presentation in an Adolescent

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Rickets can manifest with a wide variety of rheumatic symptoms. In this paper, a fifteen year old female patient with hypophosphatemic rickets presenting with symptoms suggesting sacroiliitis at disease onset is reported. The sacroiliac joint involvement in this case was attributed to the subchondral bone fractures due to the secondary hyperparathyroidism. Her symptoms resolved completely after treatment with calcitriol and phosphate solution for hypophosphatemic rickets.

Key words: hypophosphatemic rickets, sacroiliac joint

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Rickets has largely been eliminated as a medical concern even in most of the underdeveloped countries because of Vitamin D supplementation and exposure to the sun. As a result, the cases that are seen are more likely to have a genetic basis. Familial hypophosphatemic rickets, previously known as Vitamin D resistant rickets, appears to be the most common cause of inherited rickets (1).

Rickets can manifest with a wide variety of symptoms, principally with bone pain and joint involvement, that may be mistaken for a rheumatic disease (2).

We report an unusual case of hypophosphatemic rickets in an adolescent female patient. She had symptoms suggesting sacroiliac joint involvement as the first manifestation of the disease.

Case Report

A fifteen year old girl was referred to Hacettepe University Hospital, Department of Physical Medicine and Rehabilitation, with a complaint of pain in her hips and low back region. She had been in pain for 6 months and the pain had worsened gradually during the last 3 months. She also complained of fatigue and weakness and with difficulty in climbing stairs and standing up from seated position.

Physical examination revealed pain in the hip joints and low back region in the extremes of motion, pain on sacroiliac compression tests and positive Fabere test on both sides. There was a mild weakness in proximal muscles of the lower extremities (4/5 in manual muscle testing according to the Medical Research Council) and she had a waddling gait. Values for hemoglobin, white blood cell count, erythrocyte sedimentation rate, all measured with routine techniques, were normal (134 gr/L, $4.80 \times 10^3/mm^3$ and 16 mm/h respectively). Antistreptolysin-O Antibody (ASO), C-reactive protein and rheumatoid factor measured with standard serologic tests were negative. Serum alkaline phosphatase level was elevated to 2235 U/L (normal range: 70–230 U/L in 14–15 year old children). She was normocalcemic but hypophosphatemic with a serum calcium level of 2.32 mmol/L and a serum phosphorous level of 0.45 mmol/L (normal range for serum calcium and phosphorous levels are 2.12–2.62 mmol/L and 0.96–1.45 mmol/L respectively). The TmPO4/GF was 0.36 mmol/L which is far below the normal range. Renal function studies including BUN, creatinine, and renal aminoacid excretion were normal. Parathyroid hormone level, measured by specific aminoterminal radioimmunoassay was 370 pg/mL (normal range is 10–65 pg/mL). Serum 25(OH)Vitamin D_3 level was 45 nmol/L which is in the normal range of 25–100 nmol/L.

Complete skeletal X rays were obtained. X rays of the skull, hands and long bones revealed a normal bone density and appearance. However, radiological examination of the lumbosacral region demonstrated ill-defined and in some regions, sclerotic sacroiliac joints. Sacroiliac X rays confirmed the sacroiliac joint involvement (Fig. 1).

A total body radionucleide bone imaging with 15 mCi ^{99m}Tc-methylene bisphosphonate revealed diffuse radioactive uptake in all bones of the body including the mandible, pelvic bones, and the vertebrae (Fig. 2). The proximal parts of both femur and

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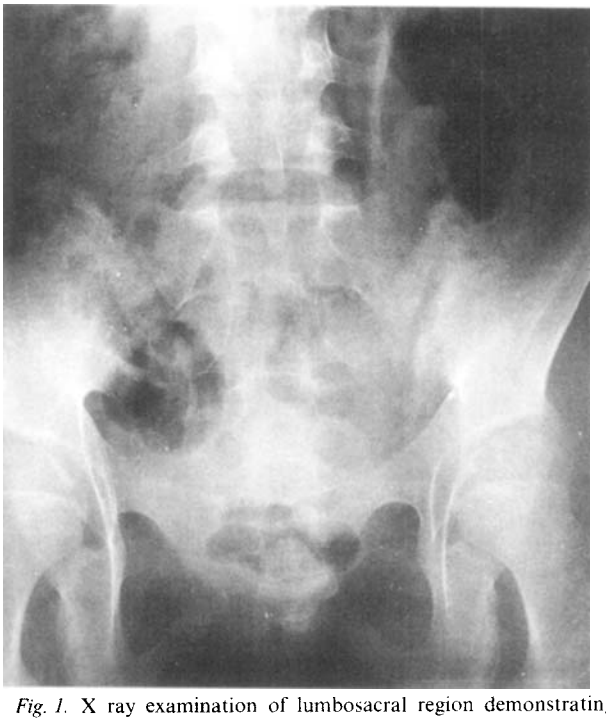

Fig. 1. X ray examination of lumbosacral region demonstrating ill-defined sacroiliac joints.

the ribs showed an irregular radioactivity. The left sacroiliac joint had a greater radioactive uptake than the right side, with an index of 1.50 and 1.28 respectively. This is above the normal accepted range on the left side (Fig. 3). As a result scintigrafic investigation suggested a metabolic bone disease and left sacroiliitis.

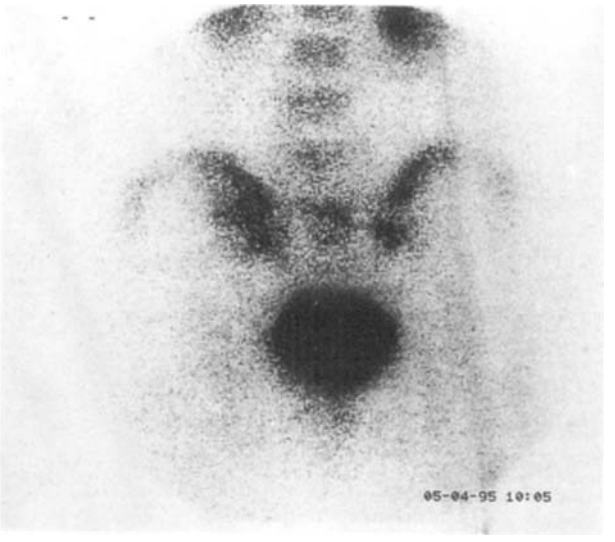

Fig. 3. Scintigraphic examination demonstrating increased radioactivity uptake in the left sacroiliac joint suggesting left sacroiliitis (posterior view).

An ultrasonographic examination of parathyroid glands was normal.

The motor and sensory nerve conduction studies and needle electromyographic investigation of the lower extremities were normal.

Although the primary complaint, physical findings, and radiological examination of the patient were consistent with sacroiliitis, the biochemical parameters and the scintigraphy suggested a metabolic bone disease as well. Normal serum calcium and 25(OH)Vitamin D_3 level, hypophosphatemia, and

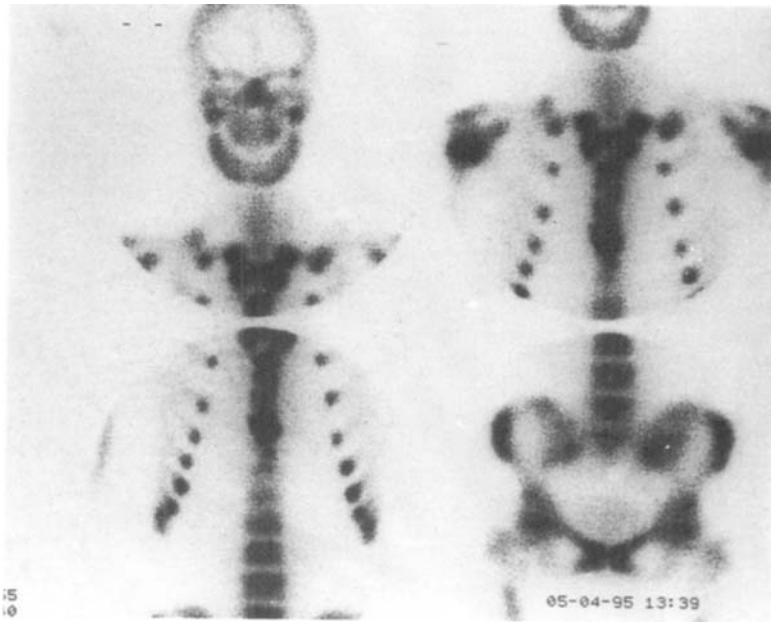

Fig. 2. Total body radionucleide imaging demonstrating diffuse radioactive uptake of all bones including the mandible, vertebrae, costochondral junctions, and the pelvis suggesting a metabolic bone disease.

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decreased renal tubular reabsorption of phosphate and secondary hyperparathyroidism strongly suggested hypophosphatemic rickets. Although bone biopsy is mandatory for the diagnosis, an attempt to obtain one was refused by the patient and her parents. Therapy was initiated with calcitriol 0.5 micrograms daily and 1 gr daily phosphate solution. After 10 weeks of treatment, she had a remarkable improvement of her pain and fatigue and her muscle weakness completely disappeared. Biochemical parameters revealed a serum calcium of 2.3 mmol/L, phosphorous of 0.6 mmol/L, and alkaline phosphatase of 1218 U/L after 10 weeks of treatment . Because of persistant hypophosphatemia the dose of calcitriol was doubled. At the end of 10 months, the patient had no complaints and biochemical studies showed a calcium of 2.32 mmol/L, phosphorous of 0.64 mmol/L and alkaline phosphotase of 741 U/L (Fig. 4). In spite of complete clinical improvement, direct sacroiliac X-ray was not significantly different at the end of 10 months of treatment. The sacroiliac joints were still ill-defined and in some regions sclerotic. However, at the end of 10 months of therapy total body radionucleide bone imaging, including the sacroiliac joints (with an index of 0.94 on the right and 0.92 on the left side) was completely normal. The improvement observed in radionucleide bone imaging confirms the diagnosis and proves the effectiveness of the treatment.

Discussion

Familial hypophosphatemic rickets has been a focus of interest since it was described by Albright and colleagues in 1937. The basis of this disorder appears to be a primary defect in phosphate transport in kidneys and intestine. The characteristic biochemical findings include hypophosphatemia due to renal phosphate wasting, normal or slightly low serum calcium, an elevated serum alkaline phosphatase, and normal or high levels of parathyroid hormone. The serum 1,25 dihydroxy Vitamin D is usually normal or slightly high, inappropriate to the low serum phosphate (2). Our patient had biochemical findings consistent with hypophosphatemic rickets. She had normal calcium, low phosphorous, and high alkaline phosphatase levels. The urinary phosphate reabsorption was definitely low despite hypophosphatemia and serum 25(OH)VitaminD_3 level was within the normal range.

Hypophosphatemic rickets shows X-linked, autosomal dominant, autosomal recessive inheritance or may be sporadic. The clinical spectrum may range from mild hypophosphatemia without any other findings, to severe bone disease with biochemical changes. In the case presented here the lack of family history and late onset of the mild symptoms without irreversible bony deformities suggest a sporadic form of the disease (2). Total body radionucleide bone imaging revealed diffuse radioactive uptake of all bones and a greater asymmetrical uptake in the left sacroiliac joint in response to the normal indices based on age and sex (3). These scintigraphic findings are most probably due to the diffuse metabolic bone disease. However, the asymmetric radioactive uptake in the left sacroiliac joint accompanied with sacroiliac joint symptoms suggests sacroiliitis. Ill-defined sacroiliac joints documented in direct X rays were also consistent with sacroiliac joint involvement, however, this appearance may be the result of a growing skeleton as expected in this age group.

Ankylosing spondylitis was also considered in differential diagnosis. However, absence of clinical signs and symptoms suggesting ankylosing spondylitis including an inflammatory type of back pain, decreased lumbar mobility, and peripheral joint involvement excluded this diagnosis.

To our knowledge, sacroiliac joint involvement associated with hypophosphatemic rickets as a first manifestation of the disease has not been reported in literature. There are some reports pointing out the involvement of axial skeleton especially in osteomalacia (4, 5). Also a recent study demonstrated calcifications of tendon and ligament insertions and joint capsules in 70% of 26 patients with X linked hypophosphatemic rickets (2). There are some reports of spinal canal stenosis secondary to hypophosphatemic Vitamin D resistant rickets (6).

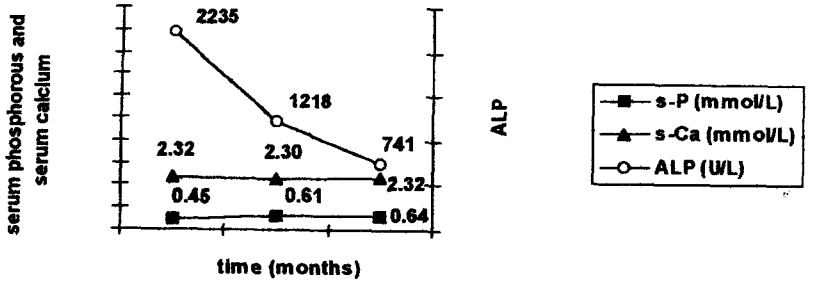

Fig. 4. Serum Alkaline Phosphatase (ALP), Calcium, and Phosphorous profile during the course of treatment

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Sacroiliac joint involvement in this patient most probably developed due to subchondral bone resorption in the sacroiliac joints because of secondary hyperparathyroidism. Subchondral bone resorption can be seen most frequently in the articulations of the axial skeleton and most distinctive at the sacroiliac joints. At sacroiliac location, radiographic findings may mimic those of ankylosing spondylitis. On roentgenograms osseous erosion and reactive new bone formation produce an ill-defined and sclerotic articular margin and pseudowidening of the joint space (5).

In conclusion, rickets may manifest with a wide variety of bone and joint symptoms which may be mistaken for a rheumatic disease. So, one should always consider metabolic diseases of bone in the differential diagnosis of diseases with articular symptoms.

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Announcements

The 3rd Pan-Arab Rheumatology Conference, 5-8 October 1997, Riyadh, Saudi Arabia.

For more information please write or email: King Faisal Specialist Hospital & Research Centre, P.Box 3354 MBC 36, Riyadh, 11211 Saudi Arabia. Tel. (966)1 4427238. Fax 4427237. Email dawn@kfshrc.edu.sa

5th RAA Congress of Rheumatology and 3rd APLAR Symposium on the Therapy of Rheumatic Diseases, 18–22 January 1998, Manila, Philippines.

Congress information: Philippine Congress Organizing Center, Physicians' Tower, 533 United Nations Avenue, Ermita, Manila 1000 Philippines. Tel. (632) 522-0541 to 48. Fax (632) 522–1090.

27th Scandinavian Congress of Rheumatology, 5–8 June 1998, Århus, Denmark

New abstract deadline: 1 March 1998. Information: Tourist Åarhus Convention, City Hall, DK–8000 Århus C, Denmark. Tel. (45)8612 1177. Fax (45)8612 0807

First International Congress on Spondyloarthropaties, 22–26 September 1998.

Organisation and secretariat: Medicongress, Waalpoel 28–34, B-9960 Assenede, Belgium. Phone (32)9 3443959. Fax (32)9 3444010. E-mail: congress@medicongress.com

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