Hypophosphataemic Rickets and Pyrophosphate Arthropathy

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Summary  Pyrophosphate arthropathy (PA) has been reported in association with a number of diseases, usually occurring in the older age group. We report a 40-year-old female with untreated X-linked hypophosphataemic rickets who presented with PA.

Key words: Hypophosphataemic Rickets, Pyrophosphate Arthropathy.

INTRODUCTION

X-linked hypophosphataemic rickets (XLH) is a familial condition characterised by inadequate mineralisation of bone, consequent skeletal abnormalities and growth retardation. It results from a renal tubular defect in phosphate transport possibly associated with abnormal vitamin D metabolism (1). Premature degenerative arthropathy is common in XLH (2). We report a patient with XLH, untreated in adult life, who presented with pyrophosphate arthropathy (PA).

CASE REPORT

A 40-year-old clerical worker with acute synovitis was referred by her general practitioner as possibly having rheumatoid arthritis. As a young child, she had been diagnosed as having rickets and treated with a vitamin D preparation from the age of 6 years to 13 years. At that time...
time she had tibial resections to straighten her legs and afterwards received no further treatment. She had first complained of pain in her knees in 1983 at the age of 33 years. At this time an X-ray was reported as showing degenerative arthritis with possible rheumatoid changes. She remained reasonably well until 9 months before attending our unit at which time she began experiencing increased pain in her knees and intermittent swelling of the right knee. Her right shoulder was painful and stiff and the right wrist intermittently painful and swollen. On examination, she was short of stature with a height of 4 foot 8 inches. There was soft tissue swelling and increased warmth over the dorsal and lateral aspect of her right wrist, but no abnormality at the shoulder. No synovial fluid was obtained on aspiration of the wrist. There was bony enlargement around the knee joints and pain and crepitus on movement of the patello-femoral joints but no effusion or soft tissue swelling.

X-rays of the hands revealed normal bone density with focal osteoporosis of both ulnar styloids, osteoarthritic changes at the first carpometacarpal joints, radio-carpal joint narrowing with sclerosis and chondrocalcinosis in the triangular ligament (Fig. 1). X-ray of the right shoulder revealed a large spur related to the infero-glenoid tubercle, some periosteal irregularity of the corocoid and of the distal end of the clavicle. X-rays of the knees showed extensive sclerosis of the right medial femoral condyle and large loose bodies in the supra-patellar pouch and popliteal fossa (Fig. 2). Tibio-femoral joint space was relatively well preserved.

Investigation revealed an inorganic phosphate level of 0.55mmol/L (NR 0.7 - 1.34) and calcium 2.42 mmol/L. Urinary calcium 2.42 mmol/L. Urinary calcium excretion was at the lower end of normal at 2.6 mmol/24 hours and urinary phosphate excretion was normal at 32.8 mmol/24 hours (NR 15 to 50). Both serum and urinary results were repeated and found to be persistent. The ESR was 25mm/1hr and the full blood count, antinuclear factor and RA latex tests were negative or normal. Although no older relative had either short stature or any form of arthritis, both the patient's daughters were under the care of the paediatricians with hypophosphataemic rickets. The patient was commenced on a phosphate preparation (Phosphate Sandoz) 2 tablets daily and 1,25 -dihydroxy cholecalciferol 500mq daily at which dose her serum phosphate normalised. During six months of follow-up, she had no further episodes of synovitis although she continued to suffer intermittent pain in her knees.

DISCUSSION

The patient described fulfills criteria for probable CPPD crystal deposition disease (3) and fits McCarty's Type C pattern of a typically female patient with low grade arthralgias and degenerative changes associated with episodic acute attacks (4). Her presentation in childhood with severe rickets unresponsive to non-hydroxylated vitamin D and persistent hypophosphataemia with inappropriate renal phosphate excretion is consistent with XLH. The X-linked dominant nature of her condition is evidenced by the fact that both her daughters have hypophosphataemic rickets despite the absence of any previous family history. She had the typical XLH radiographic findings of long bone bowing, degenerative arthropathy, extra ossicles and periosteal hyperostosis (2). The predilection for the radio-carpal compartment within the wrist demonstrated by our patient is very characteristic of PA and unlike pure degenerative joint disease (5). The triangular ligament is the commonest site of calcification to be seen in the wrist.