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Suspected Primary Distal Renal Tubular Acidosis in a Young Male with Seronegative Spondyloarthropathy, Severe Osteoporosis, Osteomalacia, and Hypothyroidism: A Case Report

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Abstract

A 22-year-old normotensive, non-diabetic male presented with recurrent episodes of hypokalemic paralysis since adolescence, followed by progressive inflammatory back pain, height loss, and proximal muscle weakness. Examination revealed severe kyphosis, restricted spinal mobility, proximal myopathy, and waddling gait. Laboratory investigations showed persistent hypokalemia (2.25-2.7 mmol/L), hyperchloraemic metabolic acidosis with normal anion gap, hypercalciuria, and elevated urinary magnesium excretion. Arterial blood gas analysis demonstrated low bicarbonate (12.5 mmol/L) with pH 7.34. Imaging confirmed bilateral sacroiliitis, medullary nephrocalcinosis, severe osteoporosis (Z score -5.5), and osteomalacia. Autoimmune screening including HLA-B27 was negative. A diagnosis of distal renal tubular acidosis (dRTA) with seronegative spondyloarthropathy, severe osteoporosis, osteomalacia, and hypothyroidism was established. The patient was managed with potassium citrate, sodium bicarbonate, sulfasalazine, teriparatide, calcium carbonate, vitamin D_3, and levothyroxine. Distal RTA is an uncommon association with seronegative spondyloarthropathy, particularly in the absence of conventional autoimmune markers. Given the adolescent onset, differentiation between primary and secondary dRTA remains a concern. Primary dRTA has been linked to mutations in ATP6V1B1, ATP6V0A4, SLC4A1, CA2, and recently FOXI1, WDR72, and ATP6V1C2. Genetic analysis is planned for this patient. This case underscores the importance of evaluating renal tubular function in young patients with recurrent hypokalemia and musculoskeletal deformities to enable early diagnosis and targeted therapy. *[J Assoc Clin Endocrinol Diabetol Bangladesh, 2025;4(Suppl 1): S51]*

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