CASE STUDIES

Renal tubular acidosis presenting as osteomalacia

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Abstract

Renal tubular acidosis (RTA) has been identified as a well-known cause of osteomalacia. The present study reporting the occurrence of osteomalacia secondary to vitamin D deficiency, underscores the need to evaluate associated RTA in patients who had pain in hip region and lower back.

Keywords: Renal tubular acidosis, osteomalacia, vitamin D

Introduction

Renal involvement, such as tubulointerstitial nephritis and renal tubular acidosis (RTA), generally occurs as an extraglandular manifestation of systemic diseases like hypergammaglobulinemia, Sjögren’s syndrome or systemic lupus erythematosus (SLE). We present here a rare sporadic case of RTA type 1 with osteomalacia.

Case Report

A 16-year-old female, born to non-consanguineous parents, presented to the clinic with a 5-year history of arthralgia and generalized body ache. She had restricted movement and pain in the hip region and lower back. Her vitals were stable and no obvious tenderness in any of the joints or any other systemic symptoms were reported. She had a waddling gait with grade 4 muscle power in all the four limbs. Her clinical investigations revealed normal blood counts, ESR and CRP. ANA by immunofluorescence was negative, but had very high levels of alkaline phosphatase (1401 IU/L). Urine analysis showed pH-7 with no protein and sugar. Urine culture was not done, as the urine routine did not show any pus cells. Levels of serum electrolytes were sodium: 131 mg/l, potassium: 3.1 mg/l, chloride: 109 mmol/dl, and bicarbonate: 14.4 mmol/l. Arterial blood gas (ABG) analysis showed pH 7.459, and PCO2 20.3 mmHg, suggestive of normal anion gap (9, normal 8-16) and mixed metabolic acidosis with respiratory alkalosis due to tachypnea. Ultrasound of abdomen showed normal kidneys without any stones. Urine osmolar gap was not done, as the patient did not show GI symptoms.

X-ray of the hands (Fig.1), pelvic region, and heels (Fig.2) showed generalized osteoporosis. Loosers zone/fracture (shown by arrows) was seen in the neck of the femur (Fig. 2). Osteomalacia was suspected, which was confirmed by elevated serum PTH level (259.8 pg/mL) and low vitamin D levels (2 ng/mL). Distil RTA was suspected as the patient’s urine was alkaline associated with hypokalemia and reduced bicarbonate levels. The inability to achieve a urine pH below 5.5, even with reduced bicarbonate, is an indication of RTA. The patient was diagnosed as having osteomalacia, vitamin D deficiency, and distil RTA. Proteinuria or glycosuria was ruled out. The patient was started on sodium bicarbonate 100 mg tablets thrice daily with vitamin D3 of 60,000 IU weekly once for 1 month, and later to monthly once dose of vitamin D3 and calcium carbonate of 500 mg daily. The patient’s alkaline phosphatase level had become normal and the waddling gait improved over a period of 6 months with normalization of serum potassium, carbon dioxide and bicarbonate on repeated arterial blood gas (ABG) test. The patient is presently on sodium bicarbonate 100 mg daily and vitamin D 60,000 IU once in 2 months along with calcium supplement.

Discussion

In the current case, osteomalacia was secondary to vitamin D deficiency and secondary hyperparathyroidism (probably nutritional). But the persistence of high urine pH
in a normal anion gap, compensated metabolic acidosis, hypokalemia and absence of proteinuria/glycosuria clearly indicated the existence of distil RTA. We speculate that the severe manifestation of osteomalacia could be the result of decrease in bone mineralization due to vitamin D deficiency and distal RTA. The co-existence of vitamin D deficiency and RTA cannot be ruled out and it is often difficult to differentiate.

In most cases, type 1 RTA develop secondary to systemic disorders like hypergammaglobulinemia, Sjögren’s syndrome, chronic active hepatitis or systemic lupus erythematosus (SLE). But it can be sporadic or familial (inherited as autosomal dominant, X-linked or recessive condition). The present case can be considered as a sporadic cause of distal RTA, since other possible causes such as urological/liver disorder, autoimmune disorder (as ANA was negative), drugs, toxins and rhabdomyolysis were absent. A recent study by Veeranna et al. has reported the occurrence of a similar sporadic case of distal RTA in a

Fig. 1: X-ray of hands

![X-ray of hands](image1)

Fig. 2: X-ray of heels and pelvis joint

![X-ray of heels and pelvis joint](image2)
4-year-old male child. The study has also highlighted on the clinical presentation of RTA as osteomalacia or rickets and renal calculi or nephrocalcinosis.¹

The study by Domrongkitchaiporn et al. has concluded that the chronic metabolic acidosis may cause suppression of bone formation and resorption in distal RTA patients, which in turn may lead to reduced bone mass. Bone acts as a buffer to neutralize the systemic acidosis as the renal impairment fails to do so. However, the researchers have reported osteomalacia as an uncommon histologic finding in such patients.²

The common causes of osteomalacia are vitamin D deficiency, certain surgeries, celiac disease, kidney or liver disorders and the use of some drugs. There are several case studies reporting the development of osteomalacia secondary to renal tubular acidosis due to Sjögren’s syndrome.³ A 2004 survey by Fulop and co-workers, based on the analysis of 250 patient records, has concluded that the occurrence of osteomalacia in patients with type 1 RTA need not be associated with Sjögren’s syndrome.⁴ This finding concurs with the present case.

It is important to identify associated RTA because only vitamin D supplementation with calcium may not be sufficient to improve the patient’s bone mineralization. Metabolic acidosis needs to be corrected with regular sodium bicarbonate supplementation, if the acquired causes are absent. An earlier study by Richards et al. has reported that osteomalacia in patients with renal tubular acidosis can be treated with sodium bicarbonate alone, owing to the inherent adverse effects of vitamin D treatment.⁵

The present study underscores the need for evaluating sporadic cause of distal RTA in patients with osteomalacia secondary to vitamin D deficiency.

Competing interests
The authors declare that they have no competing interests.

Citation

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