

## Renal Tubular Acidosis-An Unusual Presentation

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**Abstract:** A 4 year male child born to 3RD degree consanguineous parents presented with fever and loss of head control since morning, the loss of head control was sudden and was associated with pain. Preliminary examination revealed hypokalaemia and ABG revealed metabolic acidosis with normal anion gap. Urinary studies suggested pH is 6.0, urinary electrolytes suggested normal anion gap. Calcium creatinine ratio was high (1402.4mg/g) creat. Renal tubular acidosis (RTA) is a constellation of syndromes arising from different derangements of tubular acid transport. The case reported here has features of distal renal tubular acidosis with normal anion gap metabolic acidosis and urinary normal anion gap.

**Keywords:** RTA (RENAL TUBULAR ACIDOSIS)

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### I. Introduction

Distal RTA (RTA type I) is a rare renal disorder characterized by non-anion gap hyperchloremic acidosis and hypokalemia. In this condition, the alpha intercalated cells of the cortical collecting duct of the distal nephron fail to secrete acid into the urine. This failure of acid secretion leads to an inability to acidify the urine to a pH <5.5. In 1946, Albright et al. described dRTA as a distinct entity.<sup>1</sup> The clinical syndrome described, consists of hypokalemia, hyperchloremic metabolic acidosis, inability to lower urine pH below 5.5 in the face of systemic acidosis, nephrocalcinosis and nephrolithiasis. Because renal excretion is the primary means of eliminating acid from the body, there is consequently a tendency towards systemic acidemia. This leads to the clinical features of RTA type I, which include: Normal anion gap hyperchloremic metabolic acidosis; Hypokalemia (from multiple mechanisms, but often severe during periods of stress); Nephrocalcinosis; Nephrolithiasis (related to an inability to acidify urine); Hypercalciuria, and low urinary citrate); Loss of calcium from bones (which can cause rickets in children and osteomalacia in adults).<sup>2</sup>

RTA type I is either inherited or acquired. Inherited RTA type I can be either autosomal-dominant or autosomal-recessive. Autosomal-recessive RTA type I often presents in infancy, whereas autosomal-dominant RTA type I may not present until adolescence or young adulthood.<sup>3</sup> Some patients with autosomal recessive distal RTA have associated sensorineural hearing loss.<sup>4</sup> Mutations in the genes encoding carbonic anhydrase (CA) II, kidney anion exchanger-1 (kAE1), and subunits of the H<sup>+</sup>-ATPase have been identified in patients with distal RTA.<sup>5</sup> Some genetic disorders, such as Ehler-Danlos syndrome, or Wilson's disease, have also been associated with RTA type I.<sup>6</sup> In the acquired form, the disorder can be caused by drugs, autoimmune diseases, or by infection.<sup>6</sup> Some of the more common acquired forms are caused by Sjögren syndrome, lupus, hepatitis, treatment with amphotericin B, toluene toxicity, and chronic pyelonephritis.<sup>7,8</sup>

We describe a 4 year male who came with complain of fever and loss of head control since morning. Further investigations revealed hypokalaemia, Metabolic acidosis with normal anion gap and urinary normal anion gap and calcium creat ratio high. The child was managed symptomatically and was advised SHOHL's solution and gradually improved.

### II. Case Report

A 4 year old boy with fever since one day and loss of head control since morning. The loss of head control was sudden in onset and associated with pain in the neck. The child was malnourished with the weight and height below 3<sup>rd</sup> percentile and there were signs of rickets and pallor. On examination, limbs were hypotonic with absent reflexes and a motor power of grade one in all limbs. There was no history of dysarthria, diplopia, respiratory difficulty or bladder and bowel involvement. Her serum sodium was 129 mEq/L (135-148), serum potassium 1.8 mEq/L (3.5-4.5), serum chloride 111 mEq/L (95-105), with ECG changes of hypokalemia. Intravenous therapy followed by oral potassium treatment brought serum potassium to 3.3 mEq/L. Serum calcium profile revealed a value of 7.6 mg/dL. Arterial blood gas analysis showed normal anion gap (14 mEq/L) metabolic acidosis with a pH of 7.28 (7.35-7.45), serum bicarbonate was 14 mEq/L. Blood sugar, renal and liver function tests were within normal limits. Urinary anion gap was positive (3.3 mEq/L) urinary sodium 67mmol/l and chloride 64 mmol/l indicating decreased ammonium chloride secretion. Urine examination did not show any glucose, proteins or pus cells. Urinary calcium excretion was high urinary calcium/creatinine ratio was 1402.4mg/g. Child was put on alkali therapy in the form of soda bicarbonate and polycitra solution along with calcium supplementation. After two years of therapy child is now asymptomatic, with serum sodium of 141 mEq/L, potassium of 3.9 mEq/L, chloride of 103 mEq/L.

### III. Discussion

RTA is a syndrome characterized by hyperchloremic metabolic acidosis with a normal anion gap secondary to an abnormality in renal acidification as observed in the present case. In distal RTA (type 1) the distal nephron does not lower urine pH either because the collecting ducts permit excessive back diffusion of hydrogen ions from lumen to blood or because there is inadequate transport of hydrogen ions. Ammonium excretion and potassium conservation are also impaired. Majority of cases of type 1 RTA are secondary to a systemic disorder such as Sjogren's syndrome, hypergammaglobulinemia, chronic active hepatitis or systemic lupus erythematosus (SLE). But it can be sporadic or familial, inherited as autosomal dominant, X-linked and autosomal recessive condition. Absence of bicarbonaturia and fanconi syndrome distinguishes distal RTA from proximal (type 2) RTA. Clinically it may present as osteomalacia or rickets and renal calculi or nephrocalcinosis. In our case, we did not get other possible cause of secondary distal RTA like liver disorder, drug, toxins, urological disorders, rhabdomyolysis, which suggests the possibility of primary (hereditary) or sporadic cause of distal RTA in this patient, Recurrent hypokalemic paralysis with apparently progressive symptoms should be evaluated for an underlying disorder such as dRTA that could not only be potentially treatable but also achievable in growth velocity, adequate pubertal development and adult height near that of the normal parental height.

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