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DISTAL RENAL TUBULAR ACIDOSIS WITH SEVERE HYPOKALEMIA – A CASE REPORT

Paediatrics

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ABSTRACT

Background: Renal tubular acidosis is a group of transport defects secondary to either reduced distal secretion of hydrogen ion or decreased proximal tubular reabsorption of bicarbonate or both, resulting in impaired acidification of urine with persistent hyperchloremic metabolic acidosis. **Clinical description and Investigations:** Our case is that of a 5-year-old female child presenting with failure to thrive and weakness. Physical examination revealed skeletal deformities with enamel defects. Subsequent blood tests and arterial blood gas analysis revealed hyperchloremic metabolic acidosis with hypokalemia. Urine analysis revealed a positive urinary anion gap with hypercalciuria and hypocitraturia. Imaging studies revealed skeletal changes secondary to rickets and USG whole abdomen showed bilateral kidney nephrocalcinosis. Oral ammonium chloride loading was given and blood and urine pH were monitored every hour for the next 6 hours but the urine pH failed to fall below 5.3 which led us to a diagnosis of distal renal tubular acidosis. **Treatment:** Bicarbonate and potassium citrate supplementation was given along with vitamin D and calcium to this child. **Conclusion:** Timely diagnosis and initiation of appropriate supplementation can help in improving the quality of life of these patients.

KEYWORDS

distal renal tubular acidosis, hyperchloremic metabolic acidosis, hypokalemia

CASE REPORT

A 5-year-old, female child presented with failure to gain height since past 4 years along with weakness for 5 days. She also had features of pneumonia which did not resolve with conventional management and after subsequent consultation with the department of chest medicine a diagnosis of pulmonary tuberculosis was made and patient was started on anti-tubercular drugs. She had a history of multiple such episodes of fatigue at home which resolved spontaneously as per her mother. She was born of non-consanguineous marriage and was the first child of her parents. Parents gave a positive history of polyuria, polydipsia and savory for salty food items. On physical examination, she had bilateral lower limb deformity and could not stand without support on her own. She also had enamel defects (Fig. 1) and prominence of costochondral junctions. Anthropometric measurements revealed a weight of 8 kg, a height of 84 cm both of them being below 3rd percentile for age as per WHO growth charts.

Her serum sodium 139mEq/L, serum potassium 1.3 mEq/L, and arterial blood gas analysis showed pH 7.22 (7.35-7.45), bicarbonate – 20.1 mmol/L. Serum chloride levels were 106mg/dl (95-105). Serum calcium levels were 7.24mg/dl. Liver and renal function tests and blood sugar were within normal limits.

Urine output was 4.8ml/kg/hr. Urinary pH was 6.5 with no features of proteinuria, glycosuria, or hematuria. Urinary electrolytes were analyzed with urinary potassium being 10mEq/L and a positive urinary anion gap (+10) which indicates a decreased ammonium chloride secretion. 24-hour urinary citrate levels were done which revealed hypocitraturia with a level of 0.18mmol/L (1.3-6.04). 24-hour urinary calcium was 103mg/dl (0.5-35.70) which was suggestive of hypercalciuria.

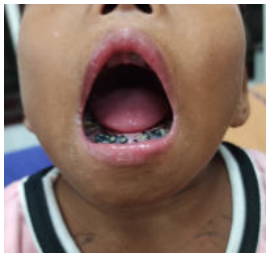

Fig 1 showing the enamel and dental defects in the patient

Digital X ray of bilateral wrist joints revealed cupping, fraying and splaying in the metaphyseal area (Fig. 2). Subsequent 25-OH Vit D3 levels were analyzed (10.97ng/ml) which revealed vitamin D deficiency. USG of the whole abdomen and KUB was done which was suggestive of nephrocalcinosis in bilateral kidneys (Fig. 3)

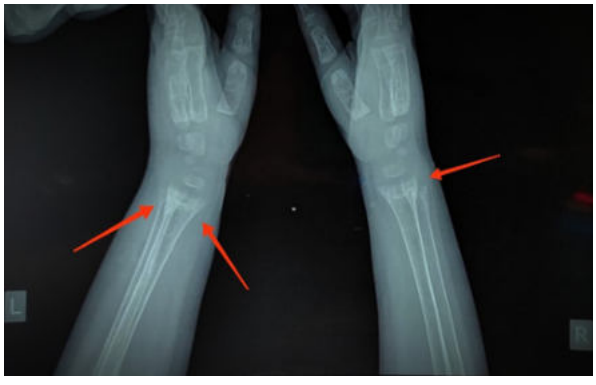

Fig 2. Digital X-Ray of bilateral wrist joints showing cupping, fraying and splaying of the metaphyseal region (marked by arrows)3

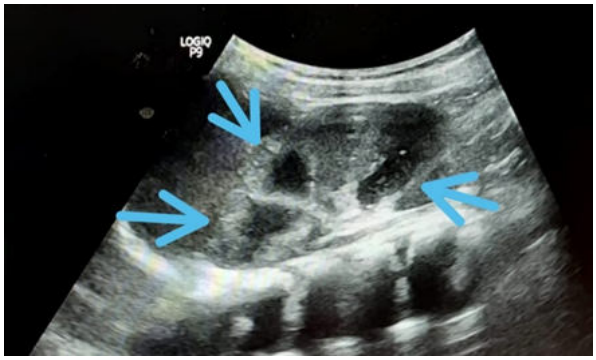

Fig 3. USG abdomen showing nephrocalcinosis in the left kidney (marked by arrows)

Diagnosis of distal renal tubular acidosis was made based on hypokalemic, hyperchloremic metabolic acidosis with normal anion gap, urinary pH of 6.5 despite systemic acidemia with hypercalciuria, hypocitraturia, bone deformities and nephrocalcinosis. Ammonium chloride test was done by giving it orally at a dose of 0.1mg/kg orally after obtaining blood pH, urine pH and serum bicarbonate. Lack of acidification of urine after six hours despite ammonium chloride therapy suggested distal RTA.

MANAGEMENT AND OUTCOME

The child was treated with sodium bicarbonate tablets, potassium citrate supplementation along with Vitamin D (as per STOSS therapy) and calcium supplementation for the illness. In subsequent follow-up after 8 weeks now the child is asymptomatic with normalization of serum potassium(4.4mEq/L) and bicarbonate levels(23mEq/L) with an arterial pH of 7.37. Subsequent digital X-ray of bilateral wrist joints showed resolution of the characteristic signs of rickets.

DISCUSSION

The term renal tubular acidosis (RTA) is applied to a group of transport defects in the reabsorption of bicarbonate (HCO_3^-), the excretion of hydrogen ions (H^+), or both which result in impaired acidification of urine. The RTA syndromes are characterized by a relatively normal GFR and a metabolic acidosis accompanied by hyperchloremia and a normal plasma anion gap [1]. There are various types of renal tubular acidosis but our case is a child with distal or type 1 renal tubular acidosis which is the most common type of RTA. Distal RTA prevalence for 2017 was estimated between 0.46 (recorded cases, of which 22.1% were considered primary) and 1.60 when including suspected cases (7.6% primary) per 10,000 people. [2] The primary or inherited form of distal RTA is mostly diagnosed in infancy, childhood, or young adulthood, while the acquired secondary form, as a consequence of other disorders or medications, can happen at any age, although it is more commonly seen in adults [3]. In distal RTA (also known as classical or type 1 RTA), there is a defect in excreting H+ ions in the distal tubule, leading to an alkaline urinary pH with calcium deposition. Diagnosis of dRTA can be challenging, since it requires a high index of suspicion and/or measurement of urinary pH after an acid load, usually in the form of oral ammonium chloride which should normally acidify the urine to pH below 5.3. [4]

In our patient, the laboratory results revealed metabolic acidosis, hypocitraturia, hypokalemia, and hypercalciuria which were consistent with those of distal renal tubular acidosis and imaging studies (USG whole abdomen with KUB) revealed nephrocalcinosis though there was no evidence of nephrolithiasis. The reason for reporting this case was that children with short stature and bone deformities with recurrent episodes of weakness and fatigue should also be evaluated for renal tubular acidosis with a high level of suspicion. Early diagnosis with the initiation of supplements and medications can improve the long-term outcome in these patients.

Review of literature was done via PubMed AND Google Scholar and very little Indian data was found in the form of case reports or case series on distal RTA though some international data was found amongst varying age groups regarding the same. Vasquez-Rios, G., Westrich, D.J., Philip, I., et al. described a case of a 57-year-old Caucasian woman with distal renal tubular acidosis. Additional laboratory workup and renal biopsy led to the diagnosis of primary Sjögren's syndrome with associated acute tubulointerstitial nephritis [5]. Abdallah JB, Charfeddine B, Braham I et al. described a 4-year-old with polyuria-polydipsia syndrome, a failure to thrive, nephrolithiasis, hypercalciuria who was subsequently diagnosed as a case of distal RTA [6]. Ranawaka, Randula et al. reported a child with distal (type 1) renal tubular acidosis presenting with progressive gross motor developmental regression and acute paralysis [7].

The current modality of treatment in these patients is by alkali/ bicarbonate supplementation at a dose of 2-4mEq/kg/24 hours along with potassium supplementation in the form of potassium citrate which helps in correcting growth, reduces calcium loss from urine and also decreases inappropriate urinary potassium losses.

CONCLUSION

Distal renal tubular acidosis is a rare disorder affecting the normal acidification of urine. It is associated with metabolic acidosis, hypokalemia, hypercalciuria, hypocitraturia, defects of bone mineralization leading to skeletal deformities and failure to thrive. A child presenting with recurrent history of weakness and fatigue with growth impairment should be thoroughly evaluated for renal tubular acidosis at the earliest hint of suspicion. Parents should be adequately counselled regarding the disease and the need for lifelong supplementation and regular follow-up. Timely diagnosis with appropriate supplementation can improve the quality of life of such patients

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DECLARATION OF CONSENT – Proper informed consent has been taken from the legal guardians of the patient regarding the publication of the information.

CONFLICT OF INTERESTS - None

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