

Rickets Secondary to Distal Renal Tubular Acidosis: A Rare Case Report.

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ABSTRACT

Rickets is a common condition affecting the paediatric patients in developing countries including India. It is usually secondary to nutritional deficiency of vitamin D. Children are predisposed to this condition if the dietary requirements of vitamin D are not met. This type of rickets is called nutritional rickets and it can be corrected by supplementation of vitamin D and calcium. But there are some other rare types of rickets which are seen secondary to renal defects. These include distal renal tubular acidosis, proximal renal tubular acidosis, vitamin D dependent rickets and hypophosphatemic rickets. The children suffering from rickets secondary to these disorders may not respond to usual doses of calcium and vitamin D. Proper diagnosis and management is required in these cases. We here present a case of 3 year old male child who presented to us with delayed walking, failure to thrive, irritability, recurrent episodes of vomitings, abdominal pain and dehydration. The patient eventually was found to be having rickets secondary to distal renal tubular acidosis.

Keywords: Distal Renal Tubular Acidosis, Rickets, Normal Anion Gap.

INTRODUCTION

Renal tubular acidosis is caused by impairment of tubular acid transport in kidney. The characteristic features of renal tubular acidosis include normal anion gap and hyperchloremic metabolic acidosis.^[1] There are 3 types of renal tubular acidosis. Distal RTA (Type I), Proximal RTA (Type II), and aldosterone insensitivity (Type IV). Distal RTA is characterized by failure of H⁺ secretion from distal nephron.^[2] This inability to secrete H⁺ ions is primarily responsible for inability to acidify the urine to acid PH. Since renal excretion is the primary mechanism by which the body excretes H⁺ ions, impaired H⁺ excretion leads to acidosis. The concurrent impaired reabsorption of potassium leads to classical metabolic derangement ie acidosis and hypokalemia. Other features which are usually seen in distal renal tubular acidosis include Normal anion gap, Hypocalcaemia, hyperchloremia, urolithiasis and rickets in children.^[3] The cause of rickets in children with distal renal tubular acidosis is bone resorption due to chronic acidosis and reduced absorption of calcium from renal tubules.^[4] This is primarily responsible for rickets due to loss of calcium. This increase calcium loss through urine may cause nephrocalcinosis and nephrolithiasis.^[5]

CASE REPORT

A 3 year old boy was admitted in our hospital with lower respiratory tract infection. He had a history of cough cold and fever since 8 days. The signs and symptoms gradually worsened and the patient developed rapid breathing and inability to take proper food since 3-4 days. On admission the baby was having respiratory distress in the form of intercostal and subcostal retractions, nasal flaring and grunting. In view of respiratory distress he was admitted in pediatric intensive care unit. A detailed history was taken. He was 2nd by order of birth. Antenatal, natal and postnatal history was uneventful. He was immunised properly. There was a significant history of delayed developmental milestones. Baby had started walking with support at around 2 years of age. According to mother he lagged behind his elder sister in achieving developmental milestones specially the gross motor milestones.

His investigations showed neutrophilic leucocytosis. Chest X-ray was suggestive of right upper lobe pneumonitis. He was initially kept Nill by mouth and oxygen inhalation was started along with IV antibiotics and IV fluids. Baby responded well to the treatment and his respiratory distress settled down and baby started taking oral feeds well. By Day 5 of admission he was shifted to ward.

During general examination he was found to be having widened wrists and other signs of rickets including frontal bossing and double malleoli

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(Marfan's sign). In view of these features X-Ray of both wrists was taken which showed classical signs of rickets including osteopenia, cupping and splaying of metaphysis [Figure 1].

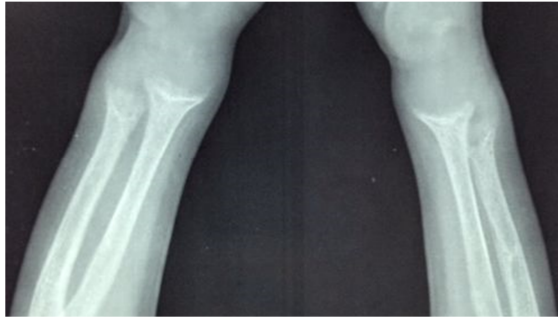


Figure 1: X-Ray both wrists showing classical features of rickets in the form of osteopenia, cupping and splaying of metaphysis.

He was worked up for rickets and the laboratory findings were found to be suggestive of rickets secondary to distal renal tubular acidosis [Table 1].

Table 1: Investigations showed hypokalemia and hyperchloraemic metabolic acidosis.

Test	Value
Serum Sodium (meq/L)	137
Serum Potassium (meq/L)	2.7
Serum Chloride (meq/L)	118
Calcium (mg/dl)	8
Phosphorus (mg/dl)	1.2
Alkaline Phosphatase (IU/lit)	2478
Urine PH	7.6
25-OH vitamin D (ng/ml)	14.20

Urine PH more than 5.5 in presence of hyperchloraemic metabolic acidosis is virtually diagnostic of distal renal tubular acidosis. Since the patient had all the classical features of distal renal tubular acidosis a diagnosis of rickets secondary to distal renal tubular acidosis was made and the patient was treated oral calcium supplementation, vitamin D. Intravenous potassium chloride replacement was given to correct hypokalemia. After correction of hypokalemia and metabolic acidosis he was discharged with an advice to remain under regular follow up.

DISCUSSION

Rickets is one of the common disorders seen in growing children. It is usually caused by deficiency of vitamin D and calcium. The common causes of vitamin D deficiency include decreased vitamin D synthesis as seen in those children who are not sufficiently exposed to sunlight for any reason. It is more prevalent in dark skinned children.^[6] Another cause of vitamin D is insufficient dietary intake of vitamin D. Strict vegetarians are at a higher risk of developing this type of nutritional rickets. Other common causes are breast fed children of vitamin D

deficient mothers, malabsorption syndromes and intake of drugs like isoniazid and glucocorticoids. While the deficiency of nutritional rickets can be corrected by supplementation of vitamin D and calcium, there are some other forms of rickets which cannot be treated by simple supplementation of calcium and Vitamin D. One of such condition is distal renal tubular acidosis.^[7]

The primary pathology in distal tubular acidosis is inability of distal tubule to secrete H⁺ ion which causes hyperchloraemic metabolic acidosis. Distal renal tubular acidosis is usually caused by Mutations in the gene that encodes AE1.^[8] Sometimes this renal tubular acidosis may be present along with progressive sensorineural deafness. In these cases the disease is inherited in autosomal recessive fashion and the mutations responsible are of ATP6V1B1 or ATPV0A4 genes.^[9] The diagnosis is usually straight forward if there is presence of hyperchloraemic metabolic acidosis and urine PH more than 5.5. Hypokalemia may also be present. All the patients having hyperchloraemic metabolic acidosis in presence of urine PH of more than 5.5 should also be investigated for sensorineural deafness and rickets.

The treatment of distal renal tubular acidosis is correction of primary metabolic abnormalities ie hyperchloraemic metabolic acidosis and hypokalemia. The secondary abnormalities like rickets, urolithiasis and skeletal abnormalities should also be appropriately managed. The correction of acidosis involves supplementation of oral potassium or sodium citrate. The hypokalemia should be managed by potassium supplementation and a potassium sparing diuretic like spironolactone, triamterene or amiloride.^[10]

Rickets seen in distal tubular acidosis can be treated by calcium and vitamin D supplementation along with correction of primary metabolic abnormality (acidosis and hypokalemia).

CONCLUSION

Rickets is a common condition affecting the pediatric patients. The most common cause of rickets is nutritional. In some rare cases rickets may be secondary to disorders like renal tubular acidosis, hypophosphatemic rickets or familial vitamin D resistant rickets. In all patients not responding to usual doses of calcium and vitamin D further tests should be conducted to rule out these conditions.

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