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Case Report

Refractory rickets in distal renal tubular acidosis: a case report of severe skeletal deformity and short stature with genetic implications

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ABSTRACT

The inability of the distal renal tubule to expel hydrogen ions is linked to type I (distal) renal tubular acidosis (RTA). Hyperchloremic metabolic acidosis, an unusual rise in urine pH, decreased excretion of ammonium and bicarbonate ions in the urine, and a slight decline in renal function are its hallmarks. Hypercalciuria is common in distal RTA because of bone resorption, which increases as a buffer against metabolic acidosis. This can result in intractable rickets. All the known genetic causes of distal RTA accounts to disfunction of intercalated cells in collecting tubules. We describe a case of refractory rickets with severe skeletal deformity and short stature secondary to distal renal tubular acidosis with probable genetic cause, who with simple intervention of alkali therapy, on follow up showed with biochemical and clinical improvement.

Keywords: Refractory rickets, Renal tubular acidosis, Nephrocalcinosis

INTRODUCTION

Unmineralized matrix at the development plates causes rickets, a disorder of growing bone. Although distal renal tubular acidosis (RTA) and other conditions that interfere with bone mineralization can cause rickets, vitamin D deficiency is the most common cause of the disease. Calcium shortage is frequently caused by hypercalciuria and nephrocalcinosis, which accompany distal RTA.[1]

Insufficient net acid excretion by the kidneys causes distal RTA, a disorder of faulty urine acidification. When systemic metabolic acidosis is present, the condition is typified by alkaline urine.[2] In its most severe form, individuals have significant hypokalemia, volume depletion, and extreme acidoticity, but their renal excretory function is generally normal. Calcium and phosphate levels are often normal, but growth is stunted, and rickets is common in instances that go untreated due to minerals draining from the bones in an effort to buffer the acidosis. Acidosis increases osteoclasts' resorption of bone. Authors report a case of distal RTA who presented with short stature, genu valgum, FTT, difficulty in walking with severe skeletal deformities.

CASE REPORT

A 5-year-old child born of 3rd degree consanguineous marriage, was admitted to the hospital due to difficulty in walking, failure to thrive, genu valgum deformity and weakness in both legs with severe pain (disturbing his sleep). On examination the child had beading over the costochondral junction and widening of the wrist joint. Ultrasonography kidney suggestive of dense echogenic shadows are seen predominantly at the tip of echogenic pyramids of bilateral kidneys medullary calcinosis. He weighed 2.5 kg when he was delivered by normal vaginal delivery at 36.2 weeks gestation. His legs were thin when he was four years old, and he had trouble walking. History of death of one male sibling at day 4 of life, cause unknown. The child received symptomatic treatment for rickets at multiple OPD visits from different private practitioners in the form of vitamin D3 sachets and syrup calcium. His vital indicators were within

normal range. At 78 cm (less than the third percentile) and 9.5 kg (less than the third percentile), he was short stature, which was proportionate in nature.

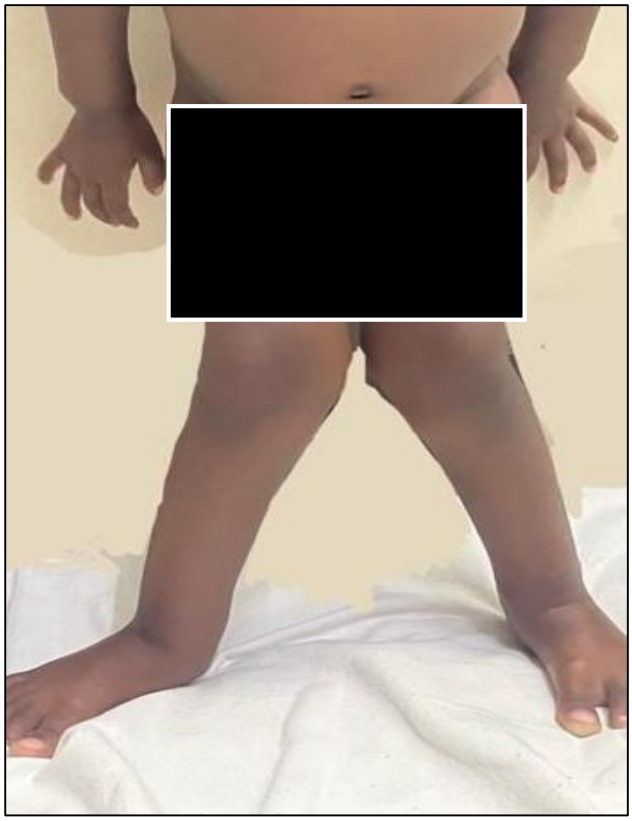

Figure 1: Clinical photograph of Genu valgum in the patient.

Figure 2: Clinical photograph wrist widening.

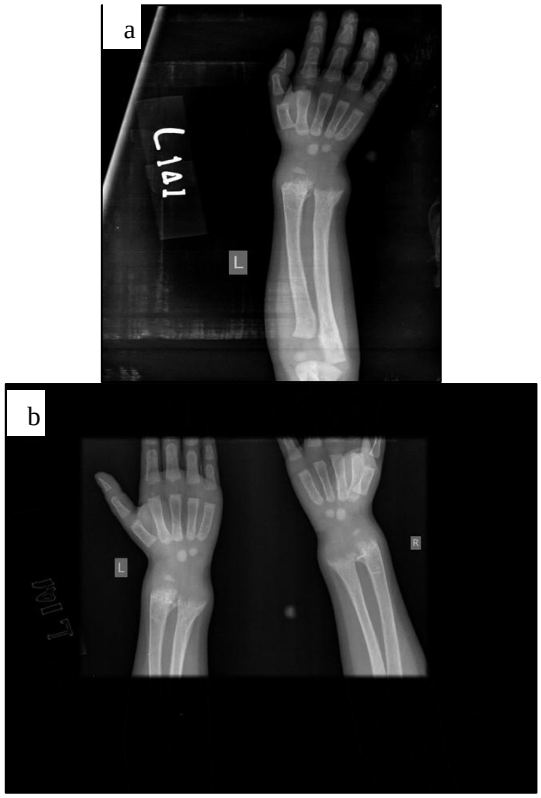

Figure 3 (a and b): Radiograph showing cupping, fraying and flaying at the wrist joint.

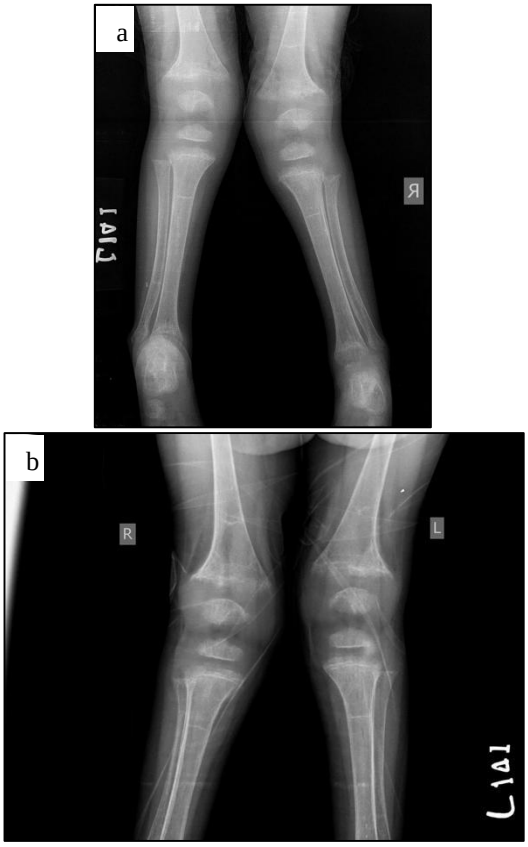

Figure 4 (a and b): Radiograph of cupping, fraying and flaying at the knee joint.

The total number of cells in the blood was normal. Hyperchloremia and hypokalemia were detected in the electrolyte battery. Alkaline phosphatase was significantly elevated, serum phosphate was reduced, and serum calcium was within the normal range. Analysis of arterial blood gas revealed a normal anion gap and mild metabolic acidosis. A positive urine anion gap, significant hypercalciuria in 24-hour urine, and reduced chloride excretion were all observed in urine electrolytes. Test results for thyroid function were normal. 25-hydroxy (25-OH) vitamin D levels were of insufficient range. Urine calcium creatinine ratio was >0.2 suggesting hypercalciuria. The levels of parathyroid hormone were significantly reduced 5.7 pg/ml. A skeletal examination revealed fraying, cupping and splaying of distal humerus and radius and widespread osteopenia. As skeletal deformity is severe, patient is planned for corrective surgery. Hearing assessment done. BERA recording showed evidence of Vth wave formation at 60 dB bilaterally. The child was treated with oral potassium citrate(2-3meq/kg/day), calcium, and vitamin D. He was discharged after a few days after his metabolic acidosis and hypokalemia were resolved. He was admitted to the hospital once more for follow-up. Pain had significantly reduced and the patient was able to sleep at night. Potassium and chloride were detected in the normal range. Alkaline phosphatase levels have decreased from before. Analysis of arterial blood gas revealed a normal anion gap and metabolic acidosis. A skeletal survey revealed cupping, flaying and splaying with development of white line of Frenkel. Urine calcium/creatinine ratio improved became <0.2.

Table 1: Illustrating biochemical investigations before and after alkali therapy.

Date	6 June 25	7 June 25	10 June 25	After treatment	16 June 25	3 July 25
Creatinine	0.38					
Sodium	139		138		139	
Potassium	3.38		4.67		4.30	
Calcium serum	8.46		8.95		8.35	
Phosphate (inorganic)	2.89				2.90	
Urine creatinine		13.64				
Urine sodium		73				
Calcium urine		4.26				
Urine potassium		20.53				
Urine chloride		133.13				
Urine pH		6.98				
Urine phosphate		13.86				
Vit D 25 OH			27.25			
CRP Quantitative	13.21					
Protein	6.07				5.74	
Albumin	3.20				3.60	
Globulin	2.87				2.14	
Bilirubin (T)	0.75				0.63	
Bilirubin (C)	0.23				0.20	
ALT	13				10	
AST	32				33	
Alk phosphatase	1168				1049	
Urea	18					
Creatinine	0.37					
A B G						
pH	7.32		7.29		7.36	7.22
pCO_2	17		21.6		27.7	32.8

DISCUSSION

RTA is a clinical illness caused by a malfunction of urine acidification that results in hyperchloremic metabolic acidosis. Acid-base exchange transporters in intercalated cells are the main factor responsible for the acidification of urine in the distal tubule. Three mechanisms are involved: cytosolic carbonic anhydrase II (CA II) provides H+ and HCO3- ions from H2O and CO2, vacuolar H+-ATPase excretes H+ ions into the collecting tubule, and the HCO3-/Cl- anion exchanger (AE1) excretes HCO3- ions into the blood. Urine acidification defects can result from impaired function of any of these components, and the significance of the impacted component determines how severe the functional fault is. Recent research has identified genetic abnormalities that affect the components' functionality. For example, studies have linked cerebral calcification and mixed-type distal

and proximal RTA to mutations in the gene producing cytosolic CA II. Distal RTA is typically the result of mutations in the AE1-encoding gene.[3] Inherited distal RTA and sensorineural hearing loss can result from mutations in the ATP6V0A4 and ATP6V1B1 genes, which encode the a4 and B1 subunits of vacuolar H+-ATPase. Since increased K+ secretion compensates for the inability to eliminate H+, most individuals with distal RTA have hypokalemia.[4,5]

The patient in this instance exhibited nephrocalcinosis, hypokalemia, alkaline urine with a positive urine anion gap, a normal blood anion gap, and persistent hyperchloremic metabolic acidosis.[6] Even though these results are in line with distal RTA, neither cerebral calcification nor sensorineural hearing loss were observed. Due to the mild metabolic acidosis, the current case also seems to be an incomplete distal RTA. To confirm the diagnosis, a genetic analysis should have been performed but due to financial constraint we were not able to perform it. This can be a study constraint.[7]

In addition to treating biochemical abnormalities, the goal of distal RTA treatment for children is to promote growth, avoid kidney stones, and prevent skeletal deformities. Alkali replacement is the cornerstone of the treatment. Because citrate salt can prevent nephrolithiasis and treat hypocitraturia, potassium or sodium citrate is better than bicarbonate.[8] Vitamin D and calcium must be replaced if rickets is present along with the distal RTA.[9,10]

CONCLUSION

His small stature could be the result of inadequate treatment or another factor. In distal RTA, some publications have noted slower development velocity and delayed bone age. The clinical characteristics of vitamin D-resistant rickets with RTA may be improved by phosphate supplementation. Therefore, thorough history and testing should be done to identify the cause of the short stature in cases that do not improve with calcium and vitamin D replacement therapy. Additionally, growth hormone and bisphosphonate medication should be considered if necessary.

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