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Nephrocalcinosis in Renal Tubular Acidosis

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ABSTRACT

Renal tubular acidosis (RTA) is a kind of hyperchloremic anion gap acidosis which occurs due to defect in urine acidification. We report an early unusual clinical presentation of Distal Renal Tubular acidosis with Nephrocalcinosis in a four months old male child, who presented with Rickets.

Keyword: *Renal Tubular Acidosis, Nephrocalcinosis, Rickets.*

INTRODUCTION

Renal tubular acidosis (RTA) is a medical condition that involves an accumulation of acid in the body due to a failure of the kidneys to appropriately acidify the urine[1]. Without proper treatment, chronic acidity of the blood leads to growth retardation, kidney stones, bone disease, chronic kidney disease, and possibly total kidney failure[2].

It is characterised by metabolic acidosis resulting from either impaired bicarbonate reabsorption or impaired hydrogen ion excretion. There are 4 types - proximal (type 2), distal (type 1) and hyperkalemic type (type 4) and Mixed[2]. Type 1 RTA is severely affected, presenting with either acute illness or growth failure at a young age. In contrast, dominant Type 1 RTA is a milder disease, and sometimes goes undiagnosed until adulthood[3].

We report an unusual clinical presentation of Distal Renal Tubular acidosis with Nephrocalcinosis in a four months old male child, who presented with bowing of leg associated with swelling & tenderness.

CASE REPORT

A four months old male child presented with complaints of bowing of left leg associated with painful swelling just above the ankle on lateral side of same leg of a size 5cm x 3cm, which was

hard in consistency and tender. There was no restriction of movement of the left ankle. Initial orthopaedic examination, vital parameters and systemic examination were normal. X-rays of lower limbs showed features suggestive of Rickets (Fig. 1), Investigations reaveled normal CBC, Calcium-10mg/dl, Phosphorus-4.3mg/dl and raised serum alkaline phosphatase-762U/l

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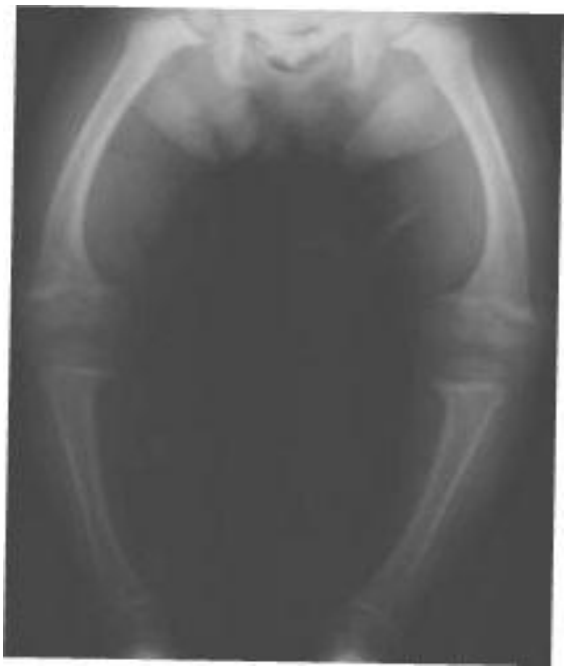

Fig. 1 – Showing Features of Rickets

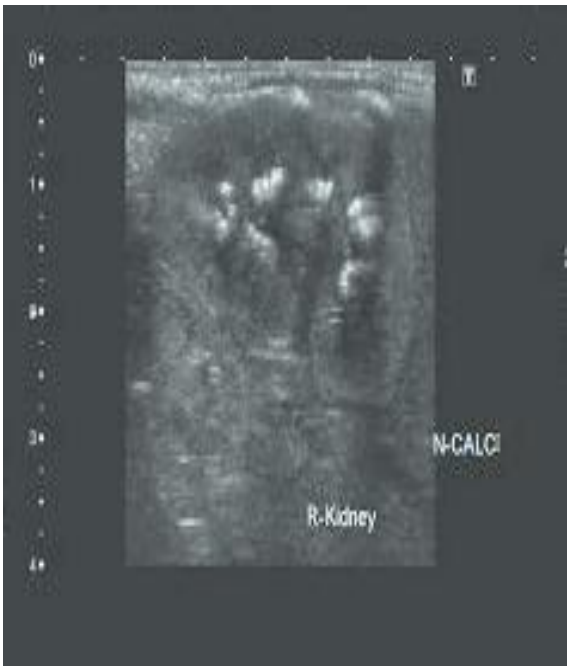

Fig. 2 – USG Showing Rt Kidney Nephrocalcinisis

A diagnosis of Rickets due to type 1 renal tubular Acidosis was made. The diagnosis of distal renal tubular acidosis was made by the findings of systemic acidosis, low bicarbonate, hypokalemia, a normal anion gap and relatively alkaline urine despite the acidemia. Nephrocalcinosis was seen on USG in the present case (Fig. 2). The patient was started on ShohlÊs solution (1mmol of sodium citrate and 1 mmol of citric acid) in a dose of 1mmol/kg/day in divided doses and vitamin D and calcium supplements result in marked improvement in swelling of leg and growth. Serum calcium, potassium and phosphorus were normalised.

DISCUSSION

To our knowledge, this is the first case report of Type 1 renal tubular acidosis with nephrocalcinosis with clinical presentation like rickets in four month old child in Indian literature.

The peculiarity of this case is early development of nephrocalcinosis with rickets in the absence of family history.

Renal Tubular Acidosis commonly presents with polyuria, constipation and failure to thrive. Proximal RTA occurs due to decreased reabsorption of filtered bicarbonate in the proximal convoluted tubule while Distal RTA is due to the defective secretion of hydrogen ions in the collecting tubules and Hyperkalemic type is due to hypoaldosteronisim or decreased

responsiveness to aldosterone which acts on the collecting tubules and causes hydrogen ion and potassium excretion[4]. Distal RTA is associated with hypocitrateuria, hypercalciuria and nephrocalcinosis.

Nephrocalcinosis is an uncommon condition in childhood where commonest cause is renal tubular acidosis, although this may not manifest itself radiographically until adolescence[4]. But some cases occurring as early as first month of life have been reported[4] similar to present child where Nephrocalcinosis is visible on USG which is rare presentation in four month child.

Distal RTA usually presents with polyuria, dehydration, anorexia, vomiting and constipation or in cases of Distal RTA secondary to interstitial nephritis/obstructive uropathy/ VUR they present with the symptoms of the disease causing the RTA[4].Our child presented with features suggestive of rickets.

Challa et al and Hanna et al have shown that metabolic acidosis directly inhibited growth hormone secretion and gene expression at target sites, thus contributing to growth failure of metabolic acidosis[5,6] . Therefore early detection ant treatment of RTA is essential for optimal growth.

Our patient was started with sodamint tablets after which swelling and tenderness reduced, subsequent blood gases revealed improvement in acidosis followed by clinical improvement.

The present case was treated with ShohlÊs solution which was well tolerated with significant improvement in growth,swelling and tenderness of legs and metabolic parameters for the one year follow up.

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