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Distal Rta Type I In Beta Thalassemia Trait With Rachitic Manifestation: A Case Report

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Abstract: *Beta thalassemia trait is a variant of beta thalassemia, which is a heterozygous state with features of mild anaemia, hypochromic microcytic red cells and elevated HbA_2.Distal renal tubular acidosis (type I RTA) is an important cause of rachitic deformities, failure to thrive and hypokalemia.It ischaracterised by persistent severe metabolic acidosis. The proximal tubular reabsorption of bicarbonate is normal. Persistant acidosis and hypercalciuria leads to nephrocalcinosis.*

Keywords*: Beta thalassemia trait, Distal renal tubular acidosis (type I), Failure to thrive,Nephrocalcinosis, Rachitic deformities.*

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I. Introduction

Thalassemia is a hereditary anaemia resulting from defect in haemoglobin production. Thalassemia refers to a spectrum of diseases characterized by reduced or absent production of one or more globin chains. In normal subjects, globin chain synthesis is very tightly controlled such that the ratio of production of alpha tonon-alpha chains is 1.00 ± 0.05. Beta thalassemiais due to impaired production of beta globin chains, leading to a relative excess of alpha globin chains, thus disrupting this ratio.Excess alpha globin chains are unstable, incapable of forming soluble tetramers on theirown, and precipitate within the cell, leading to a variety of clinical manifestations. The degree of alpha globin chain excess determines the severity of subsequent clinical manifestations, which are profound in patients homozygous for impaired beta globin synthesis and much less pronounced in heterozygotes, who generally have minimal or mild anaemia and no symptoms. The terms beta thalassemia minor or betathalassemia trait are used to describe heterozygotes, who carry one normal beta globin allele and one beta thalassemic allele. The vast majority of these patients are entirely asymptomatic, but do present an abnormal blood picture that is sometimes erroneously diagnosed as iron deficiency anaemia. However, as a rule, the microcytosis is much more profound, and the anaemia much milder, than that seen in iron deficiency anaemia. Studies of renal involvement in thalassemia syndromes have been varied and few. Renal tubular function abnormalities are well described in beta thalassemia major[3], alpha thalassemiaas well as in betathalassemia/ HbE disease.

Renal tubular acidosis comprises a group of tubular transport defects as characterised by inability to appropriately acidify the urine with resultant metabolic acidosis. Underlying abnormalities consist of an impairment of bicarbonate reabsorption or excretion of H^+ ions or combination of both occurring in the absence of or out of proportion to impairment in glomerular filtrationrate. Distal RTA(Type I) is a variant of RTA characterised by persistent severe metabolic acidosis due to impaired capacity of distal nephron to achieve a steep H^+ ion gradient between the tubular cell and lumen ,resulting in markedly decrease net secretion of H^+ions, leading to inappropriately alkaline urine[1].

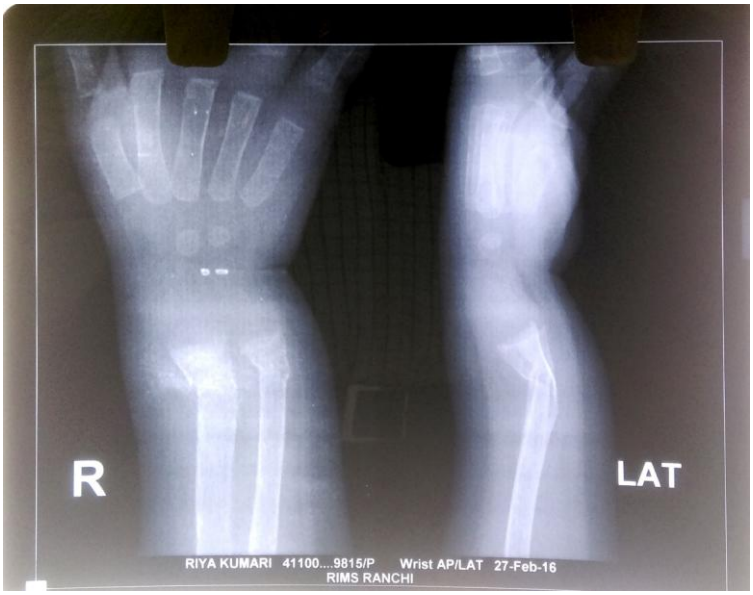
The proximal tubular absorption of bicarbonate is normal .Persistent acidosis and hypercalciuria lead to nephrocalcinosis. Distal RTA is an important cause of rachitic deformities, failure to thrive and hypokalaemia in children.[1]

II. Case Report

A 3 year old female child presented to our paediatric OPD with complaints of visible right hand deformity from 1 year, not gaining weight 6 month, abdominal distention from 4 months ,progressive pallor 3 months, polyuria& polydipsia from 2 months, fever& cough 3 days.O/E severe pallor was present, visible right hand and wrist non tender deformity, nohepatosplenomegaly, weight<50 percentile(as WHO CHART for weight for age).On further routine laboratory investigations following relevant reports were available.

ABG
•PH-7.26(7.35-7.45) •PCO_2-18.2mmhg(32-45) •PO_2-194.8mmhg(75-100) •NA-139MMOL/L(134-146) •K^+- 2.11 mmol/l(3.4-4.5) •CL^--120mmol/l(96-108) •HCT- 25%(34-52) •HCO_3-8.1MMOL/L(15-20) •ANION GAP-13.8meq/(8-16)

CBC
•HB- 5g/dl (11-13) •RBC-2.05laks/mm^3(4-5) •TLC-12,000 cells/mm^2(4000-11000) •DLC-(N-37.6%,L-52.7%,M-8.5%,E-0.7%,B-.5%) •RETIC-0.7%(0.6-1.2) • SICKLING -Negative •PBS-Predominantly hypochromic microcytic rbc with moderate anisocytosis,no immature cells seen.



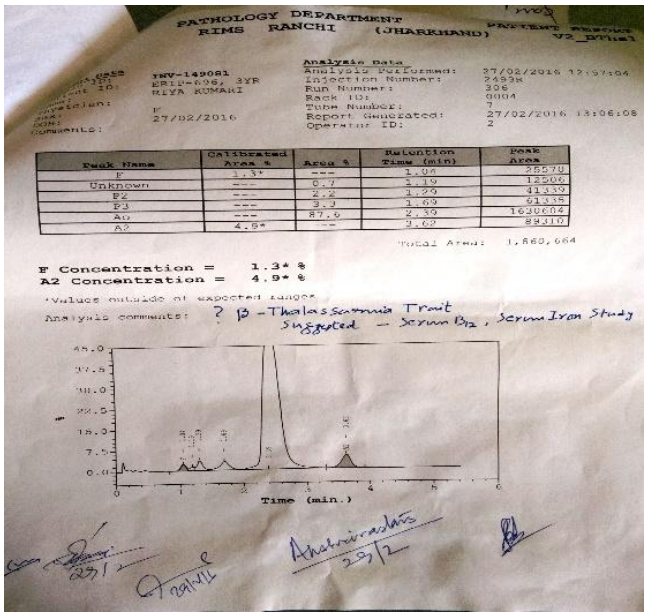
X RAY WRIST (RIGHT SIDE)

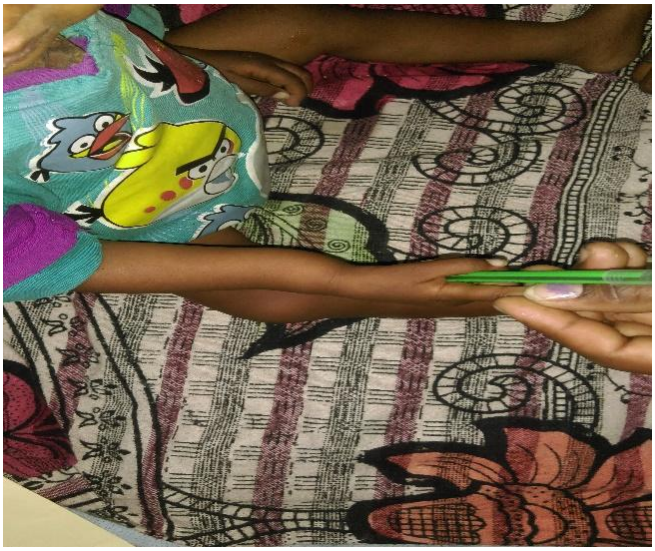
OTHER PAREMETERS

NORMAL VALUE	**PATIENT VALUE**
Blood urea(15-45mg/dl)	16
Serum creatinine(0.3-0.7mg/dl)	0.3
Total protein(6-8g/dl)	6
Serum albumin(3.5-5.5g/dl)	3.5
Serum Ca(8.8-10.6mg/dl)	5.8
Serum vitamin D(25 hydroxy vit D)-31-100ng/ml	20.2
Routine urine	Normal
Hiv/Hbsg	Negative
24hr urine creatinine(600-800mg/24hr)	15.3
Urine PH(4.6-8)	6.5
Urine specific gravity(1.003-1.030)	1.010
24hr urine k^+(25-125meq/l)	12.6
Spot Na urine(40-220mmol/l)	55
Urine CL spot test	38
Urinary anion gap(30-35meq/l)	29.6
Urine spot Ca(mg/kg/day)	7.35
Urinary calcium: creatinine ratio	0.48
Usg abdomen	Normal

HPLC suggestive of BETA THALASSEMIA TRAIT

Patient value	
HbF	1.3%
HbA2	4.9%
HbA	87.6%





RIGHT WRIST SHOWING BONE DEFORMITIES

III. Discussion

Renal tubular acidosis is a disease state characterised by normal anion gap metabolic acidosis in the setting of normal or near normal glomerular filtration rate. Thereare 4types: proximal (type II)RTA,Classic distal (type I)RTA, hyperkalaemia (type IV) RTA,and combined proximal and distal (type III) RTA[2].

In a search of English literature, we could find only three previous reports on renal tubular dysfunction in beta thalassemia minor[4,5,6]although it is well reported in beta thalassemia major[3], alpha thalassemia as well as in betathalassemia/Hb E disease. Persons with beta thalassemia minor usually are asymptomatic. Beta-thalassemia minor is characterized by bothmicrocytosis and hypochromia. It requires no treatment. Oktenli C et al first reported renal tubular dysfunction in a 20 year-old patientwith beta-thalassemia minor[5].

Kalman S et al, investigated thirty-two children with beta-thalassemia minor. The patients were classified as anaemic (haemoglobin (Hb) ≤ 11 g/dL) and non-anaemic (Hb > 11 g/dL). A control group was formed with eighteen healthy children whose ages and sexes matched those in the other groups. Fractional excretion of sodium, fractional excretion of magnesium, fractional excretion of uric acid, and tubular phosphorus reabsorption were calculated. Urinary excretion of calcium and zinc, glucosuria, beta-2 microglobulin and N-acetyl-beta-D-glycosaminidase were measured. There was no statistically significant difference among the three groups in terms of the results of any one of the above mentioned measurements[6].

Our report has some potential limitations. We have not done genetic testing to confirm thalassemiatrait and our diagnosis of thalassemia minor is based on haemoglobin electrophoresis. On electrophoresis in patients with beta thalassemia minor, over 90% of the haemoglobin will be haemoglobin-A along with an elevation in the hemoglobin-A2 value, sometimes to levels as high as 7 to 8%. We have not performed specific tests for tubular dysfunction like LDH, N-acetyl beta glycosaminidase measurementor urine electrophoresis.

Regarding, management patient was managed with oral potassium solution followed by correction metabolic acidosis with sodium bicarbonate along treatment of rachitic manifestation.

IV. Conclusion

To conclude, renal tubular dysfunction is not rare in patients with beta thalassemia minor.However, large scale studies are needed to reveal whether there is an association between

References

[1]. Pediatric nephrology ,Arvind Bagga 5th edition page no 302,303
[2]. Nelson Textbook of paediatrics ,20th edition ,page 2529
[3]. Aldudak B, Karabay Bayazit A, Noyan A, et al. Renal function in paediatric patients with beta-thalassemia major. Pediatr Nephrol 2000;15(1-2):109-12.
[4]. Cetin T, Oktenli C, Ozgurtas T, et al. Renaltubular dysfunction in beta-thalassemia minor.Am J Kidney Dis 2003;42(6):1164-8.
[5]. Oktenli C, Bulucu F. Renal tubular dysfunction in a patient with beta-thalassemia minor. Nephron 2002;92(1):222-3
[6]. Kalman S, Atay AA, Sakallioglu O, et al. Renal tubular function in children with betathalassemia minor. Nephrology (Carlton) 2005;10(5):427-9.
[7]. Cetin T, Oktenli C, Ozgurtas T, et al. Renal tubular dysfunction in beta-thalassemia minor. Am J Kidney Dis 2003;42(6):1164-8.

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