A CASE OF DISTAL RENAL TUBULAR ACIDOSIS PRESENTING AS BONE AND JOINT PAINS

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INTRODUCTION

Clinical presentation of distal Renal Tubular Acidoss is variable ranging from asymptomatic to severe growth failure, muscle weakness, stone formation, rickets, osteomalacia, renal failure and even death. Growth failure due to chronic acidosis is the commonest presentation of distal RTA in children while rickets is most common in proximal RTA[1]. We report a young girl who presented with severe bone and joint pains but physical growth parameters were above 50th percentile.

CASE REPORT

A 13 years old girl presented with generalized bone and joint pains for last five years. Complaints had progressed to difficulty in walking. Now she was severly incapacitated, non ambulatory and had to discontinue schooling a year ago. She was born of consanguineous marriage and had seven siblings. One elder brother died at age of 16years due to renal failure. Her height and weight were at 50th percentile. There was no swelling or deformities of joints, however she had generalized bone tenderness. Power was normal in all limbs. She had severe pain on walking and was restricted to bed. Rest of general and physical examination was unremarkable. There were no features suggestive of liver disease, malabsorption, Sjogren's syndrome, systemic lupus erythematosis, rheumatoid arthritis. She was receiving multiple analgesics and vitamin supplements but without relief.

Biochemical and urine examination profile is summarized in table.

Skeletal survey revealed generalized reduced bone density, intact joint spaces, widening of growth plates, multiple looser's zones, deformed pelvis, subchondral and subperiosteal bone resorption and bilateral nephrocalcinosis (Fig.1). Ultrasound abdomen showed normal size kidneys with bilateral medullary nephrocalcinosis (Fig.2).

On the basis of hyperchloromic metabolic acidosis with normal anion gap, elevated urine pH, hypercalciuria and nephrocalcinosis, she was diagnosed as a case of distal RTA.To confirm further, oral ammonium chloride test was performed. In distal RTA distal renal tubules are unable to secrete ammonium inspite of acidosis. Ammonium chloride (0.1mg/kg) was given for 3 days. In the end tests revealed worsening of acidosis, urinary pH remained above 6.0 and negative urinary anion gap,thus confirming RTA type-1. Markedly raised levels of PTH, low serum Ca^{++}, normal phosphate raised alkaline phosphatase levels but normal level of Vit.D were suggestive of secondary hyperparathyroidism due to marked hypercalciuria leading to marked bone resorption resulting in tenderness and severe incapacitating state.

She was started with alkali treatment (Bicarbonate 2 meq/kg/day). After about 01month, blood pH was 7.37, $HCO3^-$ 21meq /L,S. urea 8.3mmol/L,$S.K^+$ 4.2mmol /l,$S.Na^+$ 139mmol/l, alkaline phosphatase 2030U/L. Treatment with chlorothiazide (2 mg/kg/day) was given for hypercalciuria. Clinical response was very encouraging. Body aches and pains were significantly relieved and she became ambulatory performing routine household activities. On follow up after 6 months PTH levels were 22.6 mol/l, alkaline phosphatase 1522U/L and she had gained 3kg of weight. Levels of blood pH, $HCO3^-$, were within normal limits and urinary calcium was 6.5mmol/24 hours (upper normal range). S.urea was 8.0 mmol/l. Her family was requested to bring other siblings for investigation but they did not comply with.

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Table: Results of Investigations

Investigations	Initial	Follow up	Normal value
Hb%(g/dl)	13.1	13.6	12-15
Na^+(meq/l)	141	140	136-149
K^+(meq/l)	4.4	4.5	3.5-5.0
Cl^- (meq/l)	109	100	98-106
Calcium(mmol/L)	1.86	2.1	2.1-2.65
Phosphate	0.95	1.2	0.8-1.65
Alkaline phosphatase(U/L)	5020	1530	Up to 645
Creatinine(umol/L)	131	110	27-62
Urea(mmol/l)	9.2	8.3	3.3-6.7
Parathyroid hormone(Pmol/L)	73.1	22.5	0.8-6.0
RA factor	negative	-	
pH	7.21	7.37	7.35-7.45
Bicarbonate(meq/L)	12	21	21-28
Base excess(meq/L)	-13	-4.8	_+5
Urine pH	6.5	6.4	<6.0
Urinary calcium(mmol/24 hrs)	15.8	6.5	1.25-6.0
Urinary phosphate(mmol/24 hrs)	10.0	-	<32.3
Vit.D3 (ng/ml)	26.93	-	>30

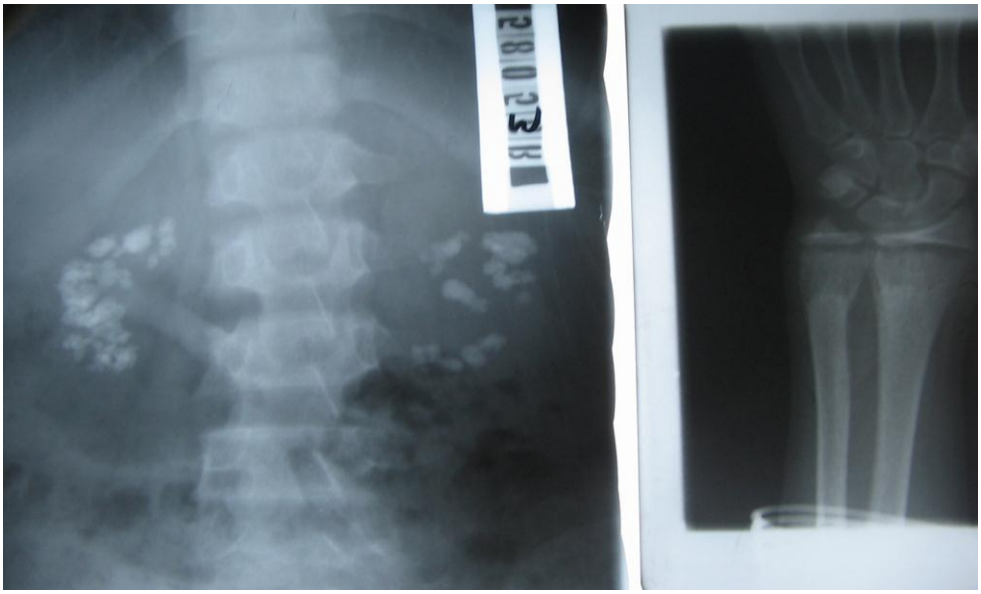

Fig.2 **Fig.1**

Regular follow up every three months and biochemical monitoring was advised and parents were explained about life long duration of treatment.

DISCUSSION

Distal RTA has diverse presentation- ranging from being completely asymptomatic to frank renal failure. It usually presents with failure to thrive, polyuria and polydipsia in children. Acidotic breathing, severe muscular weakness due to hypokalemia, renal stones and frank renal failure are more common presenting features in adults. Hyperchloremic metabolic acidosis with normal anion gap and alkaline urinary pH form major diagnostic criteria. Hypocalcaemia, hypercalciuria, raised alkaline phosphatase levels, nephrocalcinosis can be seen in patients with delayed diagnosis who also develop signs of rickets. In this

patient above mentioned findings led to diagnosis of distal RTA.

Generalized muscular weakness, sometimes even leading to respiratory failure has been reported[2]. But generalized body and joint pains with severe osteopenia due to distal RTA as in this case have been rarely reported in children. A case with severe bony deformities and multiple fractures (initially misdiagnosed as Osteogenesis Imperfecta) was reported from India in 2001[3]. Rickets is usually seen in cases of proximal RTA and only mild osteopenia is seen in distal RTA. Cause of rickets in these cases is not clear because vitamin D metabolites levels are found usually within normal range as was in this case also. Vitamin D is not required in these cases because alkali supplementation is sufficient enough leading to resolution of rachitic changes[4]. We also did not use vitamin D in this case because it can be potentially dangerous for already existing hypercalciuria.

Hypercaciuria is proposed to cause hypocalcaemia and secondary hyperparathyroidism in distal RTA. Hyperparathyroidism leads to osteopenia and subperiosteal bone resorption especially in neglected cases and those with delayed diagnosis[5]. These changes may mimic rachitic changes but are not typical of rickets. In this case also radiologiocal changes on skeletal survey were not those of typical rickets although alkaline phosphatase levels were markedly elevated. Rather changes were more of bone demineralization and resorption explaining complaints of severe body aches and bone tenderness. Oral alkali supplementation in dose of 2-3meq/kg/day is required to treat distal RTA life long. This treatment not only corrects metabolic acidosis but also other biochemical abnormalities including bony changes[6]. Thiazide diuretics are advised in cases of persistent hypercalciuria to prevent nephrocalcinosis and renal failure. Our patient showed quite satisfactory response to this treatment. Late diagnosis was reason of advanced skeletal changes leading to these complaints which could be prevented with early intervention.

REFERENCES

1. Bagga A,Bajpai A,Menon S. Approach to renal tubular disorders. Indian J Pediatr 2005;72:771-6.
2. Zakria M,Nadeem M,Dar NR. Renal tubular acidosis presenting as quadriparesis. Pak Armed Forces Med J 2003;53:249-50.
3. Bajpai A,Gulati S,Singh A,Bagga A. Distal renal tubular acidosis with severe bony deformities and multiple fractures. Indian Pediatr 2001;38:1301-5
4. Bajpai A,Bagga A,Hari P,Bardia A,Mantan M. Long term outcome in children with primary distal renal tubular acidosis. Indian Pediatr 2005;42:321-8.
5. Haider I,Alam I,Sherin A.Renal tubular acidosis: A case report. J Postgrad Med Inst 2004;18:697-700.
6. Derakshan A, Basiratnia M,Fallahzadeh MH, Hosseini-Al-Hashemi G. A Neglected case of Renal Tubular Acidosis. Saudi J Kidney Dis Transpl 2007;18:585-9.