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Case Report

Paediatrics Section

Medullary Sponge Kidney in a Paediatric Patient: A Rare Case Report

SACHIN DANGI[1], NAMITA GWASIKOTI[2], KAPIL BHALLA[3], POONAM DALAL[4]

ABSTRACT

Medullary Sponge Kidney (MSK) is a rare disease which is characterised by dilatation of the collecting ducts in one or both kidneys. These patients are more prone to develop recurrent kidney stones, nephrocalcinosis, repeated Urinary Tract Infections (UTIs) and distal Renal Tubular Acidosis (dRTA). Presentation is very uncommon in children, it usually presents at 10-30 years of life. Here we report a case of five-year-old boy who presented to us with the complaints of inadequate weight gain and short stature. Patient was consistently having hypokalemia with normal anion gap metabolic acidosis and laboratory features were suggestive of dRTA. Computed tomography of abdomen revealed bilateral nephrocalcinosis with increased medullary echogenicity consistent with MSK. Patient was started on oral alkali therapy and showed significant improvement in growth.

Keywords: Distal renal tubular acidosis, Hypokalemia, Nephrocalcinosis

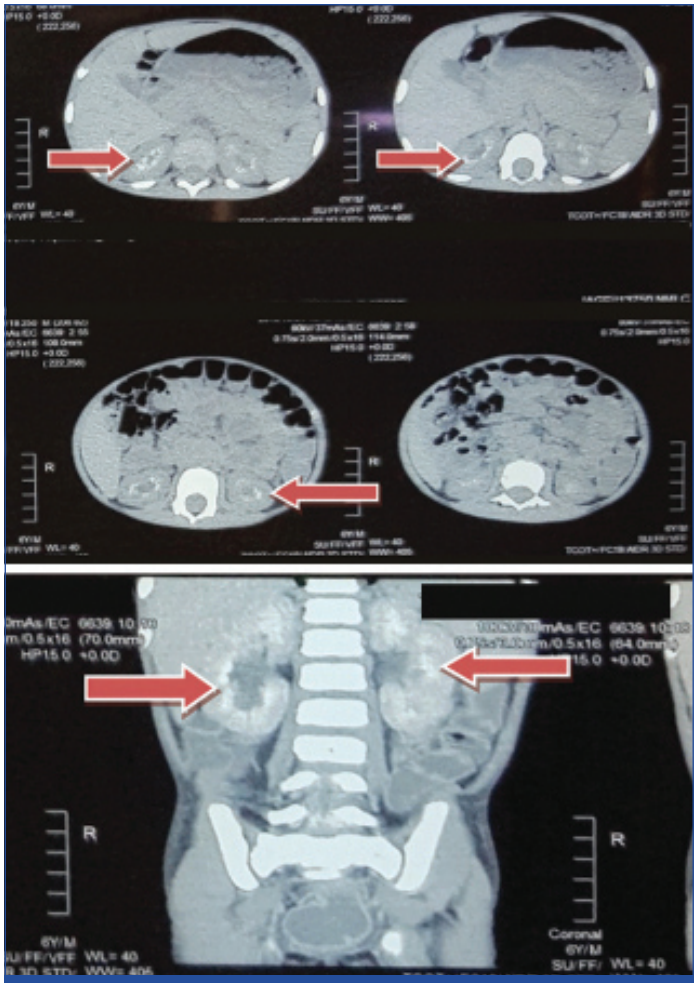
CASE REPORT

A five-year-old boy presented to our paediatrics department with the complaints of short stature and inadequate weight gain noticed by parents since last two years. There was no history of recurrent diarrhoea, vomiting, fever or any other illness. There was also no history of short stature or any other chronic illness in family members. On anthropometric examination, his height was 90 cm (< -3SD), weight was 8.8 kg (< -3 SD) and weight for height < -3 SD (Protein energy malnutrition grade IV) [1]. Physical examination revealed wrist widening and disabled gait with knock knee deformity [Table/Fig-1]. Detailed work-up for failure to thrive with short stature was done. His blood counts, liver function tests, thyroid hormone assays and echocardiography were within normal limits. Laboratory evaluation revealed hyperchloremic metabolic acidosis with a pH of 7.2 and serum bicarbonate level 11 mEq/L with normal anion gap [Table/Fig-2]. Vitamin D3 levels were 28 ng/mL and serum alkaline phosphatase 465 U/L. His serum potassium was consistently on the lower side even after corrections (<2.5 mmol/L). Patient's urinary pH was consistently above 5.5. Based on the above findings a diagnosis of dRTA was made. Ultrasonography of Kidney, Ureter and Bladder (KUB) region showed non-specific findings of bilateral raised echogenicity. We planned a CT scan of abdomen which showed bilateral medullary nephrocalcinosis with increased medullary echogenicity consistent with MSK [Table/Fig-3]. Patient was started on alkali therapy with oral sodium citrate solution at 4 mEq/kg/day. After six months of treatment, patient showed significant improvement in growth and is currently on follow-up.

[Table/Fig-1]: Five-year-old boy with short stature and knock knees.

Test	Result
Blood	
pH (venous)	7.20 (N: 7.35-7.45)
pCO_2 (venous)	17.6 mmHg (N: 32-45)
HCO_3 (venous)	11 mEq/L (N: 22-29)
Sodium	138 mmol/l (N: 135-145)
Potassium	2.6 mmol/l (N: 3.5-5)
Chloride	114 mmol/l (N: 98-106)
Uric acid	2.4 mg/dL (N: 1.7-5.8)
Calcium	9.8 mg/dL (N: 8.8-10.8)
Phosphorous	3.5 mg/dL (N: 3.7-5.6)
Alkaline phosphatase	465 U/L (N 145-420)
Serum anion gap	7
Vitamin-D	28 ng/mL
T3,T4,TSH	80, 6.5, 0.95
Haemoglobin	9.0 gmL
Total leucocyte count	9200/mm^3
Blood urea	20.1 mg/dL
Serum creatinine	0.9 mg/dL
Urine	
Specific gravity	1.010
pH	6.5
Glucose	Negative
Proteins	Normal
Calcium	12.5 mg/kg/day (N<4)
Echocardiography	Normal

[Table/Fig-2]: Laboratory evaluation of patient during hospital stay.



[Table/Fig-3]: CT abdomen showing bilateral medullary nephrocalcinosis (arrow).

DISCUSSION

The MSK is a rare developmental abnormality defined by cystic dilatations of the precalyceal collecting ducts within the medullary pyramids of one kidney or both kidneys. Its precise prevalence is unknown but has been estimated to be around 1 in 10,000 to 20,000 [2]. It is thought to be a congenital anomaly with delayed expression but familial forms have also been described. It is an uncommon entity in childhood. It is usually diagnosed at 10 to 30 years of life [3]. Although these patients can maintain normal renal functions through adulthood, they are at a high risk of complications such as nephrolithiasis, pyelonephritis, hyposthenuria (inability to concentrate urine), and distal RTA. There are only a few case reports of childhood diagnosis of this disease in already existing literature [4-6].

RTA is a diseased state characterised by normal anion gap metabolic acidosis. There are four main types of RTA, Distal (type I) RTA, proximal (type II) RTA, combined type III and hyperkalemic (type IV) RTA [7].

Distal RTA can be sporadic or inherited. It results from an impaired functioning of transporters or proteins in the distal tubules involved in acidification process. Despite the presence of severe metabolic acidosis, urine pH cannot be reduced to <5.5 due to impaired hydrogen ion excretion. To compensate decreased hydrogen ion excretion, there is an increased K+ secretion distally, which leads to hypokalemia. Distal RTA is characterised by urine pH above 5.5, hyperchloremic, hypokalemic normal anion gap metabolic acidosis, hypercalciuria, hypocitraturia, hyperkaluria, and normal renal function tests [8]. In the index case, all laboratory parameters were compatible with dRTA. Patients with dRTA present with non anion gap metabolic acidosis and growth failure [9]. Present case patient also presented with failure to thrive. Distinguishing features of dRTA from proximal RTA include nephrocalcinosis and hypercalciuria. In the index case, nephrocalcinosis was seen in bilateral kidney. Diagnosis of MSK was made by radiographic imaging. Intravenous pyelography is the gold standard for MSK diagnosis but it is not commonly performed due to prevalence of CT scan [10]. CT scan can be used to visualise renal calcification [11]. In present case, CT scan revealed medullary calcifications.

Kumar SV et al., described a case of 14-year-old male child who was admitted with complaints of failure to thrive and, growth retardation. Laboratory evaluation showed normal anion gap, hyperchloremic metabolic acidosis with respiratory compensation and persistent alkaline urine (pH >7) consistent with dRTA. USG of KUB region showed bilateral dense calcification in the calyces with distal acoustic shadow. Dilated calyces with calcification giving paint brush radiating outwards from calyces were seen on intravenous urology which was strongly suggestive of MSK [12]. In the present case, also patient presented with growth retardation with normal anion gap metabolic acidosis.

Gupta R et al., reported a case of five-year-old girl who presented with recurrent episodes of weakness of all four limbs. She had hypokalemia, polyuria, failure to thrive and metabolic acidosis. Based on clinical features and laboratory features, a diagnosis of distal renal tubular acidosis was made. Ct abdomen showed hypoechoic renal medulla [13]. In present case patient did not have complaints of limb weakness. CT abdomen of present case patient showed medullary nephrocalcinosis with raised medullary echogencity.

Outcome in MSK is usually good as long as urinary tract infections and nephrolithiasis are prevented.

CONCLUSION(S)

MSK associated with dRTA should be suspected in any child who presents with failure to thrive, persistent hypokalemia, metabolic acidosis and nephrocalcinosis. Growth failure and other metabolic disorders associated with tubular dysfunctions may improve with simple alkali therapy.

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PARTICULARS OF CONTRIBUTORS:
1. Junior Resident, Department of Paediatrics, Pt BD Sharma PGIMS, Rohtak, Haryana, India.
2. Junior Resident, Department of Paediatrics, Pt BD Sharma PGIMS, Rohtak, Haryana, India.
3. Associate Professor, Department of Paediatrics, Pt BD Sharma PGIMS, Rohtak, Haryana, India.
4. Professor, Department of Paediatrics, Pt BD Sharma PGIMS, Rohtak, Haryana, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Sachin Dangi,
Rohtak, Haryana, India.
E-mail: Sachindangi31@gmail.com

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