When to suspect Renal tubular disorder in a child

Kiran Sathe

Pediatric Nephrology

Sir H. N. Reliance Foundation Hospital, Mumbai

ECHO India

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KIDNEY FOUNDATION FOR CHILDREN

Renal tubular disorders

- Often present with subtle and non specific presentation unlike glomerular disorders
- May mimic common illnesses and hence may go unnoticed
- Often significantly affects overall physical growth of the child
- Requires repeated and specific testing to arrive at a possible diagnosis
- Slow progression to chronic kidney disease unlike glomerular disorders

Renal tubular functions

- Conserve water
- Conserve electrolytes- Na, K, Cl, Ca, P, Mg
- Acid base homeostasis
  (bicarbonate reabsorption- proximal tubule &
  acid excretion- distal tubules)
- Maintain GFR by tubular- glomerular feedback
- Erythropoietin production
- Promote normal growth
- Promote bone health
- Normal blood pressure

Common presentation of renal tubular disorders

- Antenatal presentation- polyhydramnios
- Poor growth (poor gain in weight and height)
- Developmental delay
- Repeated dehydration and fever
- Polyuria, polydipsia
- Electrolyte disturbances- Na, K
- Acid base disturbances- Metabolic acidosis and metabolic alkalosis
- Rickets, Muscle weakness
- Renal stones and nephrocalcinosis
- Hypertension
- Gradual progression to CKD, (low eGFR)

Group classification of renal tubulopathy

- Rickets- non nutritional rickets
- Metabolic alkalosis
- Metabolic acidosis- Normal gap metabolic acidosis
- Polyuria- salt wasting (RTA) or pure water loss (Diabetes insipidus)
- Renal stones
- Chronic kidney disease (CKD)

Possible aetiology based on main presentation

- **Rickets- non nutritional rickets**

- Hypophosphatemia rickets +/- proximal RTA/ Fanconi syndrome
- Vit D dependent rickets
- Renal failure associated rickets

- **Metabolic alkalosis**

- Associated with conditions causing renal chloride wasting
- Urine chloride > 40 meq/L
- Saline resistant metabolic alkalosis

- Bartter syndrome
- Gitelmans syndrome
- Hyperaldosteronism

- **Metabolic acidosis**
- Results from inadequate acid excretion by renal tubules
- Usually associated with normal GFR
- Normal anion gap metabolic acidosis

Proximal RTA

Generalised or isolated proximal tubular defect

Genetic defects in specific transport channels

Storage condition- Galactosemia, Glycogen storage disorder, tyrosinemia, Wilson disease, etc

Distal RTA

obstructive uropathy or genetic defects in transport channels

Hyperkalemic type 4 RTA

Hyperkalemia and metabolic acidosis resulting from aldosterone related defects

Polyuria- inability to concentrate urine

- Salt + water loss- RTA, Bartter syndrome

- **Pure water loss-** DI (central , nephrogenic DI)
- Genetic defects
- Secondary causes- electrolyte disturbances
- Infiltration disorders

Renal stones, nephrocalcinosis

- Proximal tubulopathy- RTA, Dents disease
- Distal RTA
- Primary Hyperoxaluria
- Hypercalcemia, Hypercalciuria
- Primary hyperparathyroidism

Chronic Kidney disease (CKD)

- All tubular disorders can progress to CKD over a variable period
- May not present with oliguria, edema unlike glomerular disorders

- Nephronopthisis, polycystic kidneys
- Chronic tubulointerstitial disease
- Drug induced, FSGS, nephrocalcinosis

Basic work up in a suspected tubulopathy

- CBC- anemia
- BUN, Creatinine, eGFR
- Serum electrolytes, Venous blood gas
- Anion gap
- Normal AG= {Na- (Cl+ HC03)}= 8-12
- Serum calcium, phosphate, uric acid, magnesium,
- 25OH vit D, 1,25 OH Vit D, PTH, alkaline phosphatase,
- X ray long bones
- USG KUB- renal sizes, CMD, echotexture, stones, obstructive uropathy, bladder
- Urine routine – pH, SG, casts, crystals, RBC, protein , leucocytes

Special tests may be done if required

- **Electrolytes imbalances**
- Hyponatremia- urine electrolytes, Fractional excretion of sodium
- Hypernatremia- water deprivation tests, ADH challenge
- Hypokalemia/ hyperkalemia-
- TTKG (to assess effect of aldosterone on renal distal nephron)

- **Rickets-** tubular reabsorption of phosphorous (TRP and TmP/ GFR)
- Normally > 85% proximal tubular reabsorption of phosphate seen
- Genetic testing if available

- **Metabolic alkalosis-**
- Urine chloride, Blood pressure
- Plasma renin, serum aldosterone
- Genetic testing if available

- **RTA-**
- Distal RTA- urine calcium: creat, hearing assessment, USG KUB, genetic test
- Proximal RTA- urine glucose, Urine protein, serum Po4, TRP, opthal evaluation
- Specialised tests as needed

Principles of management

- Often children may present with complications related to the underlying problem
- May need repeated testing to confirm diagnosis
- Prompt fluid resuscitation in patients with shock
- Appropriate replacement of electrolytes
- Correction of acid base imbalances
- Monitor eGFR in suspected tubulopathy to identify CKD especially when presents with polyuria
- Genetic diagnosis needed in unclear situations