

## Pediatric Proximal Renal Tubular Acidosis: A Clinical Approach

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An inherited or acquired clinical syndrome known as proximal renal tubular acidosis (pRTA) is characterized by normal anion gap hyperchloremic metabolic acidosis and decreased bicarbonate reclamation in the proximal tubule.

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## Methods and Materials

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**Laboratory tests:** B, ... A, ...

**Urine pH measurement:** ...

**Serum electrolyte assessment:** ...

**Renal Function tests:** B, ... (B ...)

**Kidney ultrasound:** ...

**Renal scintigraphy:** ...

## Genetic testing

Landau D (2021) Pediatric Proximal Renal Tubular Acidosis: A Clinical Approach. J Obes Metab 4: 146.

**Clinical features:**

Proximal renal tubular acidosis (PRTA) is a rare renal tubular disorder characterized by a defect in the proximal renal tubule's ability to reabsorb bicarbonate. This leads to a metabolic acidosis with a normal anion gap. The clinical presentation is often insidious, with symptoms such as muscle weakness, growth retardation, and bone disease. The diagnosis is confirmed by a urine pH > 5.5 in the presence of systemic acidosis.

**Underlying causes:**

The underlying causes of PRTA are diverse and can be categorized into genetic and acquired forms. Genetic causes include mutations in the SLC6A6 gene, which encodes the sodium-dependent bicarbonate cotransporter (NBCe1). Acquired causes include Fanconi syndrome, which is a generalized defect of proximal tubular function, and certain medications like ifosfamide.

**Diagnosis:**

The diagnosis of PRTA is based on a combination of clinical, laboratory, and genetic findings. Key laboratory features include a metabolic acidosis with a normal anion gap, a urine pH > 5.5, and a urine bicarbonate concentration > 20 mEq/L. Genetic testing for SLC6A6 mutations is available for confirmation. The differential diagnosis includes distal renal tubular acidosis (DRTA) and Fanconi syndrome.

**Treatment:**

The primary treatment for PRTA is the administration of bicarbonate salts, such as sodium bicarbonate or potassium bicarbonate. This helps to correct the metabolic acidosis and improve symptoms. In cases of Fanconi syndrome, additional treatments like phosphate and vitamin D supplements may be required. Regular monitoring of electrolytes and renal function is essential for long-term management.

**Long-term outlook:**

The long-term outlook for PRTA is generally good with appropriate treatment. Patients may experience growth retardation and bone disease, but these can be managed with bicarbonate therapy and other supportive measures. Regular follow-up with a nephrologist is recommended to monitor for complications and adjust treatment as needed.

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**Conflict of Interest**

The author declares that there is no conflict of interest regarding the publication of this article.

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