

## 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:** ... A, ...

**Serum electrolyte assessment:** ...

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

**Kidney ultrasound:** ... A.

**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 growth retardation, muscle weakness, and bone pain. Laboratory findings typically show a low serum bicarbonate level, a normal anion gap, and a urine pH that is inappropriately high for the degree of acidosis. The underlying defect is usually a deficiency of the sodium-dependent bicarbonate cotransporter (NBCe1) in the proximal tubule.

**Underlying causes:**

The primary cause of PRTA is a genetic defect in the NBCe1 gene, which encodes the sodium-dependent bicarbonate cotransporter. This defect is inherited in an autosomal recessive manner. In some cases, PRTA can be secondary to other renal tubular disorders, such as Fanconi syndrome or distal renal tubular acidosis (DRTA). The diagnosis is confirmed by genetic testing and a renal biopsy showing characteristic tubular defects.