

**DISEASE:**  
**Autosomal recessive distal renal tubular acidosis**

<b>NAME:</b>	Autosomal recessive distal renal tubular acidosis
<b>DESCRIPTION:</b>	A rare autosomal recessive form of proximal renal tubular acidosis (pRTA) characterized by an isolated defect in the proximal tubule leading to the decreased reabsorption of bicarbonate and consequently causing urinary bicarbonate wastage. Mild growth retardation and reduced bone density are extra-renal complications. Several fractures and delayed puberty are possible features.
<b>ORPHACODE:</b>	402041
<b>SYNOMYS:</b>	AR dRTA Autosomal recessive distal RTA
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	<a href="#">FOXI1</a> <a href="#">ATP6V0A4</a> <a href="#">ATP6V1B1</a> <a href="#">WDR72</a>
<b>CREATED:</b>	13 May 2019 - 01:02

**CHANGED:**

22 Jun 2023 - 16:14

---

Source URL: <http://gentest.healthdata.be/disease/2296>

## RELATED CONTENT

---

### Related Genetic Tests

- [Tubulopathy \(gene panel\)](#)

### Related Laboratories

- [Centre de Génétique-Institut de Pathologie et de Génétique \(IPG\)](#)

### Related Analytes

- [ATPase H+ transporting V0 subunit a4](#)
- [ATPase H+ transporting V1 subunit B1](#)
- [forkhead box I1](#)
- [WD repeat domain 72](#)

### Related Gene Panels

- [Tubulopathy/Nephrolithiasis \(106 genes\) - IPG](#)

---

Source URL: <http://gentest.healthdata.be/disease/2296>