

Disease

SLC4A4 associated proximal renal tubular acidosis^{1, 2}

Gene

SLC4A4

Inheritance	AR
Current Treatments	oral alkali replacement therapy, potassium chloride
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum bicarbonate, chloride, potassium, urinary pH and anion gap. ³
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC4A4&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/pubmed/24978391>

² <https://omim.org/entry/604278>

³ <https://pubmed.ncbi.nlm.nih.gov/29914390/>