

Disease

Hypokalemic tubulopathy and deafness^{1, 2}

Gene

KCNJ16

Inheritance	AR
Current Treatments	Oral bicarbonate and potassium. Indomethacin, magnesium, spironolactone in some cases.
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum bicarbonate, chloride, potassium, urinary pH and anion gap. Renin and aldosterone levels. ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=kcnj16&checkSpell=false

References

¹ <https://omim.org/entry/619406>

² <https://pubmed.ncbi.nlm.nih.gov/37466410/>