

Disease

RENI syndrome^{1, 2}

Gene

SGPL1

Inheritance	AR
Current Treatments	pyridoxine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	flow cytometric analysis, white blood cell count, cortisol level, urine analysis ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/search?cond=SGPL1

References

¹ <https://omim.org/entry/617575>

² <https://pubmed.ncbi.nlm.nih.gov/32233035/>