

Disease

Infantile-onset, biotin-responsive neurodegeneration^{1, 2}

Gene

SLC5A6

Inheritance	AR
Current Treatments	biotin, pantothenic acid, lipoate
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC5A6&term=&cntry=&state=&city=&dist=

References

¹ <https://www.omim.org/entry/618973>

² <https://pubmed.ncbi.nlm.nih.gov/31392107/>