

Disease

Brown-Vialetto-Van Laere syndrome 2¹

Gene

SLC52A2

Inheritance	AR
Current Treatments	riboflavin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma acylcarnitine profile, urine organic acids ²
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC52A2&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK299312/>

² <https://pubmed.ncbi.nlm.nih.gov/32909658/>