

Disease

Thiamine metabolism dysfunction syndrome-2^{1, 2}

Gene

SLC19A3

Inheritance	AR
Current Treatments	B1 (thiamine) and biotin
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC19A3&term=&cntry=&state=&city=&dist=

References

¹ <https://omim.org/entry/607483>

² <https://www.ncbi.nlm.nih.gov/books/NBK169615/>