

Disease

Taurine transporter deficiency¹

Gene

SLC6A6

Inheritance	AR
Current Treatments	taurine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	serum amino acids for taurine level ¹
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC6A6&term=&cntry=&state=&city=&dist=

References

¹ <https://pubmed.ncbi.nlm.nih.gov/31903486/>