

Disease

Progressive myoclonic epilepsy 4^{1, 2}

Gene

SCARB2

Inheritance	AR
Current Treatments	Miglustat
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma glucosylsphingosine ³
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=scarb2&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/254900>
- ² <https://pubmed.ncbi.nlm.nih.gov/34337151/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/34275752/>