

Disease

Early infantile epileptic encephalopathy-25^{1, 2, 3}

Gene

SLC13A5

Inheritance	AR
Current Treatments	Ketogenic diet, stiripentol
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	plasma and CSF citrate levels ⁴
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=SLC13A5&term=&cntry=&state=&city=&dist=

References

- ¹ <https://omim.org/entry/615905>
- ² <https://www.ncbi.nlm.nih.gov/pubmed/29895383>
- ³ <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9498415/>
- ⁴ <https://pubmed.ncbi.nlm.nih.gov/28673551/>