

Disease

GLUT1 deficiency syndrome-1^{1, 2}

Gene

SLC2A1

Inheritance	AR and AD
Current Treatments	ketogenic diet and carnitine, avoid barbiturates, methyxanthine(caffeine), valproic acid
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	comparison of blood glucose concentration with CSF glucose concentration obtained after 4 hr fast ¹
Disease Frequency (per 100,000)	1.2 ³
Genes Included in Disease Frequency	SLC2A1
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=glut1&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/books/NBK1430/>
- ² <https://pubmed.ncbi.nlm.nih.gov/32913944/>
- ³ <https://www.ncbi.nlm.nih.gov/pubmed/26537434>