

Disease

Early infantile epileptic encephalopathy-7^{1, 2, 3}

Gene

KCNQ2

Inheritance	AD
Current Treatments	carbamazepine, ezogabine for loss-of function variants, sodium channel blockers, pyridoxine
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	no
Disease Frequency (per 100,000)	2.8 ⁴
Genes Included in Disease Frequency	KCNQ2
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=kcnq2&term=&cntry=&state=&city=&dist=

References

- ¹ <https://www.ncbi.nlm.nih.gov/pubmed/30771507>
- ² <https://pubmed.ncbi.nlm.nih.gov/35906921/>
- ³ <https://pubmed.ncbi.nlm.nih.gov/32863083/>
- ⁴ <https://www.kcnq2cure.org/kcnq2-epilepsy/>