

**Disease**

# Early infantile epileptic encephalopathy-14<sup>1, 2, 3</sup>

**Gene**

## KCNT1

|                                                 |                                                                                                                                                                                                                           |
|-------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AD                                                                                                                                                                                                                        |
| <b>Current Treatments</b>                       | quinidine for gain of function variants                                                                                                                                                                                   |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                            |
| <b>Non-Molecular Testing Availability</b>       | no                                                                                                                                                                                                                        |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=kcnt1&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=kcnt1&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

**References**

- <sup>1</sup> <https://www.ncbi.nlm.nih.gov/pubmed/28987752>
- <sup>2</sup> <https://www.ncbi.nlm.nih.gov/pubmed/30112700>
- <sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/32505479/>