

Disease

# Early infantile epileptic encephalopathy-50<sup>1, 2, 3</sup>

Gene

**CAD**

|                                                 |                                                                                                                                                                                                                                                   |
|-------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                                                |
| <b>Current Treatments</b>                       | uridine, triacetyluridine                                                                                                                                                                                                                         |
| <b>Evidence for Treatment</b>                   | expert opinion                                                                                                                                                                                                                                    |
| <b>Non-Molecular Testing Availability</b>       | yes                                                                                                                                                                                                                                               |
| <b>Non-Molecular Test</b>                       | complete blood count and peripheral blood smear <sup>3</sup>                                                                                                                                                                                      |
| <b>Experimental / Investigational Therapies</b> | <a href="https://clinicaltrials.gov/ct2/results?cond=CAD+trifunctional&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=CAD+trifunctional&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

References

<sup>1</sup> <https://pubmed.ncbi.nlm.nih.gov/33249780/>

<sup>2</sup> <https://pubmed.ncbi.nlm.nih.gov/32820246/>

<sup>3</sup> <https://pubmed.ncbi.nlm.nih.gov/32117025/>