

**Disease**

# Early-onset progressive encephalopathy with brain edema and/or leukoencephalopathy 1<sup>1, 2</sup>

**Gene**

**NAXE**

|                                                 |                                                                                                                                                                                                                         |
|-------------------------------------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Inheritance</b>                              | AR                                                                                                                                                                                                                      |
| <b>Current Treatments</b>                       | niacin                                                                                                                                                                                                                  |
| <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=NAXE&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=">https://clinicaltrials.gov/ct2/results?cond=NAXE&amp;term=&amp;cntry=&amp;state=&amp;city=&amp;dist=</a> |

**References**

<sup>1</sup> <https://omim.org/entry/617186>

<sup>2</sup> <https://www.sciencedirect.com/science/article/abs/pii/S1096719222002992>