EARS2 gene

glutamyl-tRNA synthetase 2, mitochondrial

Normal Function

The *EARS2* gene provides instructions for making an enzyme called mitochondrial glutamyl-tRNA synthetase. This enzyme is important in the production (synthesis) of proteins in cellular structures called mitochondria, the energy-producing centers in cells. While most protein synthesis occurs in the fluid surrounding the cell nucleus (cytoplasm), some proteins are synthesized in the mitochondria.

During protein synthesis, in either the mitochondria or the cytoplasm, a type of RNA called transfer RNA (tRNA) helps assemble protein building blocks called amino acids into a chain that forms the protein. Each tRNA carries a specific amino acid to the growing chain. Enzymes called aminoacyl-tRNA synthetases, including mitochondrial glutamyl-tRNA synthetase, attach a particular amino acid to a specific tRNA. Mitochondrial glutamyl-tRNA synthetase attaches the amino acid glutamate to the correct tRNA, which helps ensure that glutamate is added at the proper place in the mitochondrial protein.

Health Conditions Related to Genetic Changes

Leukoencephalopathy with thalamus and brainstem involvement and high lactate

At least 23 mutations in the *EARS2* gene have been found in individuals with leukoencephalopathy with thalamus and brainstem involvement and high lactate (LTBL), a condition characterized by abnormalities in certain brain regions, including the thalamus and the brainstem (the part of the brain that connects to the spinal cord), and a high level of a substance called lactate in the brain and elsewhere in the body. Affected individuals typically have problems with thinking and motor abilities and with controlling muscle function.

The *EARS2* gene mutations involved in LTBL likely reduce the amount of mitochondrial glutamyl-tRNA synthetase. A shortage of this protein is thought to prevent the normal assembly of new proteins within mitochondria. Researchers speculate that impaired protein assembly disrupts mitochondrial energy production. However, it is unclear exactly how *EARS2* gene mutations lead to the features of LTBL.

Leigh syndrome

MedlinePlus Genetics provides information about Leigh syndrome

Other Names for This Gene

- COXPD12
- gluRS
- glutamate tRNA ligase 2, mitochondrial
- glutamate--tRNA ligase
- KIAA1970
- MSE1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EARS2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=124454[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28EARS2%5BTIAB%5D%29+AND+english%5Bla%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLUTAMYL-tRNA SYNTHETASE 2, MITOCHONDRIAL; EARS2 (https://omim.org/entry/612799)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/124454)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=EARS2[gene])

References

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Genomic Location

The *EARS2* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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