ETFDH gene

electron transfer flavoprotein dehydrogenase

Normal Function

The *ETFDH* gene provides instructions for making an enzyme called electron transfer flavoprotein dehydrogenase. This enzyme is normally active in the mitochondria, the energy-producing centers in cells. Electron transfer flavoprotein dehydrogenase is involved in the process by which fats and proteins are broken down to produce energy.

Health Conditions Related to Genetic Changes

Glutaric acidemia type II

Some mutations in the *ETFDH* gene prevent the production of the electron transfer flavoprotein dehydrogenase enzyme. Other mutations result in the production of a defective enzyme that cannot fulfill its role in the series of reactions (metabolic pathways) that break down fats and proteins. This enzyme deficiency allows these nutrients, as well as compounds created as the nutrients are partially broken down, to build up to abnormal levels, especially when the body is under stress. Toxic products of incomplete metabolism damage cells in many body systems, resulting in the signs and symptoms of glutaric acidemia type II.

Chromosomal Location

Cytogenetic Location: 4q32.1, which is the long (q) arm of chromosome 4 at position 32.1

Molecular Location: base pairs 158,672,101 to 158,709,623 on chromosome 4 (Homo sapiens Updated Annotation Release 109.20190905, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI
Other Names for This Gene

- electron transfer flavoprotein-Q oxidoreductases
- electron transfer flavoprotein ubiquinone oxidoreductase
- electron-transferring-flavoprotein dehydrogenase
- ETF dehydrogenase
- ETF-ubiquinone oxidoreductase
- ETFD_HUMAN
- ETFQO

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ETFDH%5BTIAB%5D+OR+%28electron-transferring-flavoprotein+dehydrogenase%5BTIAB%5D%29%29+OR+%28MADD%5BTIAB%5D%29+OR+%28ETFQO%5BTIAB%5D%29+OR+%28ETF-ubiquinone+oxidoreductase%5BTIAB%5D%29+OR+%28Electron+transfer+flavoprotein%5BTIAB%5D%29+OR+%28ubiquinone+oxidoreductase%5BTIAB%5D%29+OR+AND+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ELECTRON TRANSFER FLAVOPROTEIN DEHYDROGENASE
  http://omim.org/entry/231675

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ETFDH%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2110
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q16134
Sources for This Summary

- OMIM: ELECTRON TRANSFER FLAVOPROTEIN DEHYDROGENASE
  http://omim.org/entry/231675


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