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.

Other Names for This Gene

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

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ETFDH (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2110[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ETFDH%5BTIAB%5D%29+OR+%28electron-transferring-flavoprotein+dehydrogenase%5BTIAB%5D%29%29+OR+%28%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+%28electron+transfer+flavoprotein+ubiquinone+oxidoreductase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+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; ETFDH (https://omim.org/entry/231675)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2110)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ETFDH[gene])

References

- Goodman SI, Binard RJ, Woontner MR, Frerman FE. Glutaric acidemia type II:gene structure and mutations of the electron transfer flavoprotein: ubiquinoneoxidoreductase (ETF:QO) gene. Mol Genet Metab. 2002 Sep-Oct;77(1-2): 86-90. doi:10.1016/s1096-7192(02)00138-5. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12359134)
- Olsen RK, Andresen BS, Christensen E, Bross P, Skovby F, Gregersen N. Clearrelationship between ETF/ETFDH genotype and phenotype in patients with multipleacyl-CoA dehydrogenation deficiency. Hum Mutat. 2003 Jul;22(1):12-23. doi: 10.1002/humu.10226. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12815589)
- Olsen RK, Andresen BS, Christensen E, Mandel H, Skovby F, Nielsen JP, KnudsenI, Vianey-Saban C, Simonsen H, Gregersen N. DNA-based prenatal diagnosis forsevere and variant forms of multiple acyl-CoA dehydrogenation deficiency. PrenatDiagn. 2005 Jan;25(1):60-4. doi: 10.1002/pd.983. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15662686)
- Olsen RK, Olpin SE, Andresen BS, Miedzybrodzka ZH, Pourfarzam M, Merinero B, Frerman FE, Beresford MW, Dean JC, Cornelius N, Andersen O, Oldfors A, Holme

E,Gregersen N, Turnbull DM, Morris AA. ETFDH mutations as a major cause ofriboflavin-responsive multiple acyl-CoA dehydrogenation deficiency. Brain. 2007Aug;130(Pt 8):2045-54. doi: 10.1093/brain/awm135. Epub 2007 Jun 20. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17584774)

- Spector EB, Seltzer WK, Goodman SI. Assignment of electron transferflavoprotein-ubiquinone oxidoreductase (ETF-QO) to human chromosome 4q33 byfluorescence in situ hybridization and somatic cell hybridization. Mol GenetMetab. 1999 Aug;67(4): 364-7. doi: 10.1006/mgme.1999.2873. Citation on PubMed (https://pubmed.ncbi.nlm .nih.gov/10444348)
- White RA, Dowler LL, Angeloni SV, Koeller DM. Assignment of Etfdh, Etfb, andEtfa to chromosomes 3, 7, and 13: the mouse homologs of genes responsible forglutaric acidemia type II in human. Genomics. 1996 Apr 1;33(1):131-4. doi:10.1006/geno. 1996.0170. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8617498)

Genomic Location

The *ETFDH* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

Last updated July 1, 2008