ETFB gene
electron transfer flavoprotein subunit beta

Normal Function

The *ETFB* gene provides instructions for making one part (the beta subunit) of an enzyme called electron transfer flavoprotein. This enzyme is normally active in the mitochondria, the energy-producing centers in cells. Electron transfer flavoprotein 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 *ETFB* gene prevent the production of the electron transfer flavoprotein 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: 19q13.41, which is the long (q) arm of chromosome 19 at position 13.41

Molecular Location: base pairs 51,345,155 to 51,366,418 on chromosome 19 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)
Other Names for This Gene

- electron transfer flavoprotein beta subunit
- electron transfer flavoprotein, beta polypeptide
- electron-transfer-flavoprotein, beta polypeptide
- ETFB_HUMAN
- FP585

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28ETFB%5BTIAB%5D%29+OR+%28MADD%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+beta+subunit%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+beta-subunit%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ELECTRON TRANSFER FLAVOPROTEIN, BETA POLYPEPTIDE
  http://omim.org/entry/130410

Research Resources

- ClinVar
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=3482
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2109
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P38117
Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12706375

- OMIM: ELECTRON TRANSFER FLAVOPROTEIN, BETA POLYPEPTIDE  
  http://omim.org/entry/130410

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12815589

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15662686

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16510302

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8617498

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8688100

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