ETFA gene

electron transfer flavoprotein subunit alpha

Normal Function

The *ETFA* gene provides instructions for making one part (the alpha 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 *ETFA* 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.

Other Names for This Gene

- electron transfer flavoprotein alpha subunit
- electron transfer flavoprotein alpha-subunit
- Electron transfer flavoprotein, alpha polypeptide
- electron-transfer-flavoprotein, alpha polypeptide
- electron-transfer-flavoprotein, alpha polypeptide (glutaric aciduria II)
- electron-transferring-flavoprotein, alpha polypeptide (glutaric aciduria II)
- EMA
- ETFA_HUMAN
- GA2
- MADD

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ETFA (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2108[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28ETFA%5BTIAB%5D%29+OR+%28%28MADD%5BTIAB%5D%29+OR+%28electron+transfer+flavoprotein+alpha-subunit%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- ELECTRON TRANSFER FLAVOPROTEIN, ALPHA POLYPEPTIDE; ETFA (https://omim.org/entry/608053)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2108)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=ETFA[gene])

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Genomic Location

The *ETFA* gene is found on chromosome 15 (https://medlineplus.gov/genetics/chromosome/15/).

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