TMEM70 gene

transmembrane protein 70

Normal Function

The *TMEM70* gene provides instructions for making a protein called transmembrane protein 70. This protein is found in cell structures called mitochondria, which convert the energy from food into a form that cells can use. Transmembrane protein 70 is thought to play an important role in assembling and stabilizing a group of proteins called complex V. Complex V is the last of five complexes that carry out a multistep process called oxidative phosphorylation, through which cells derive much of their energy. Complex V is involved in the final step of oxidative phosphorylation. Specifically, one segment of complex V allows positively charged particles, called protons, to flow across a specialized membrane inside mitochondria. Another segment of complex V uses the energy created by this proton flow to convert a molecule called adenosine diphosphate (ADP) to adenosine triphosphate (ATP), which is used by the cell as energy.

Transmembrane protein 70 is also thought to be involved in the assembly of complex I, which is the first mitochondrial complex involved in oxidative phosphorylation.

Health Conditions Related to Genetic Changes

Mitochondrial complex V deficiency

At least 12 mutations in the *TMEM70* gene have been identified in people who have mitochondrial complex V deficiency, a disorder with a wide variety of signs and symptoms. A few of these gene mutations are particular to people of Roma or Arab descent, and account for the majority of mitochondrial complex V deficiency cases caused by *TMEM70* gene mutations. This disorder can also be caused by mutations in other genes.

The signs and symptoms of mitochondrial complex V deficiency are most prominent in organs and tissues that require a large amount of energy, such as the brain and heart. Abnormal brain function (encephalopathy) and other neurological problems can occur. Another common feature of mitochondrial complex V deficiency, especially when caused by *TMEM70* gene mutations, is hypertrophic cardiomyopathy. This condition is characterized by thickening (hypertrophy) of the heart (cardiac) muscle that can lead to heart failure. *TMEM70* gene mutations alter transmembrane protein 70 and impair its ability to perform its function in complex V assembly. As a result, the amount of complex

V in cells is reduced. The resulting impairment of oxidative phosphorylation and energy production leads to the signs and symptoms of mitochondrial complex V deficiency.

Other Names for This Gene

- FLJ20533
- MC5DN2
- transmembrane protein 70, mitochondrial isoform a
- transmembrane protein 70, mitochondrial isoform b

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TMEM70 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=54968[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TMEM70%5BTIAB%5D%29+OR+%28transmembrane+protein+70%5BTIAB%5D%29%29+OR+%28%28transmembrane+protein+70,+mitochondrial+isoform+a%5BTIAB%5D%29+OR+%28transmembrane+protein+70,+mitochondrial+isoform+b%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TRANSMEMBRANE PROTEIN 70; TMEM70 (https://omim.org/entry/612418)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/54968)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TMEM70[gene])

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Genomic Location

The *TMEM70* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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