TAFAZZIN gene

tafazzin, phospholipid-lysophospholipid transacylase

Normal Function

The *TAFAZZIN* gene provides instructions for producing a protein called tafazzin. Several different versions (isoforms) of the tafazzin protein are produced from the *TAFAZZIN* gene. Most isoforms are found in all tissues, but some are found only in certain types of cells. The tafazzin protein is located in structures called mitochondria, which are the energy-producing centers of cells. Tafazzin is involved in altering a fat (lipid) called cardiolipin, which plays critical roles in the mitochondrial inner membrane. The tafazzin protein adds a fatty acid called linoleic acid to the cardiolipin molecule, which enables cardiolipin to perform its functions. Cardiolipin is necessary for maintaining mitochondrial shape, energy production, and protein transport within cells.

Health Conditions Related to Genetic Changes

Barth syndrome

More than 130 mutations in the *TAFAZZIN* gene have been found to cause Barth syndrome. This rare condition occurs almost exclusively in males and is characterized by a weakened heart (cardiomyopathy), muscle weakness, recurrent infections, and short stature. *TAFAZZIN* gene mutations that cause Barth syndrome result in the production of tafazzin proteins with little or no function. As a result, linoleic acid is not added to cardiolipin, which results in a reduction of functional cardiolipin. In addition, a variant of cardiolipin called monolysocardiolipin (MLCL) is formed. A lack of functional cardiolipin and an excess of MLCL are thought to cause problems with normal mitochondrial shape and functions such as energy production and protein transport. Tissues with high energy demands, such as the heart and other muscles, are most susceptible to cell death due to reduced energy production in mitochondria. Additionally, affected white blood cells have abnormally shaped mitochondria, which could impair their ability to grow (proliferate), mature (differentiate), and function, leading to a weakened immune system and recurrent infections. Dysfunctional mitochondria likely lead to other signs and symptoms of Barth syndrome.

Familial dilated cardiomyopathy

MedlinePlus Genetics provides information about Familial dilated cardiomyopathy

Left ventricular noncompaction

MedlinePlus Genetics provides information about Left ventricular noncompaction

Other disorders

Some mutations in the *TAFAZZIN* gene cause dilated cardiomyopathy without the other features of Barth syndrome (described above). Dilated cardiomyopathy is a condition in which the heart becomes weakened and enlarged and cannot pump blood efficiently, often resulting in heart failure. The decreased blood flow can lead to swelling in the legs and abdomen, fluid in the lungs, and an increased risk of blood clots.

Mutations in the *TAFAZZIN* gene can also cause a heart condition called isolated noncompaction of left ventricular myocardium (INVM). This condition occurs when the lower left chamber of the heart (left ventricle) does not develop correctly. In INVM, the heart muscle is weakened and cannot pump blood efficiently. Abnormal heart rhythms (arrhythmias) can also occur. INVM frequently causes heart failure.

Other Names for This Gene

- BTHS
- CMD3A
- EFE
- EFE2
- G4.5
- LVNCX
- tafazzin (cardiomyopathy, dilated 3A (X-linked); endocardial fibroelastosis 2; Barth syndrome)
- TAZ_HUMAN
- XAP-2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TAFAZZIN (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6901[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28TAZ%5BTIAB%5D%29+OR+%28tafazzin%5BTIAB%5D%29+OR+%28Barth+syndrome%5BTIAB%5D%29%29+OR+%28%28BTHS%5BTIAB%5D%29+OR+%28CMD3A%5BTIAB%5D%29+OR+%28EFE%5BTIAB%5D%29+OR+%28EFE2%5BTIAB%5D%29+OR+%28G4.5%5BTIAB%5D%29+OR+%28tafazzin%5BTIAB%5D%29+OR+%28XAP-2%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TAFAZZIN, PHOSPHOLIPID-LYSOPHOSPHOLIPID TRANSACYLASE; TAFAZZIN (https://omim.org/entry/300394)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6901)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TAZ[gene])

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Genomic Location

The *TAFAZZIN* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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