MPV17 gene

mitochondrial inner membrane protein MPV17

Normal Function

The *MPV17* gene provides instructions for making a protein whose function is largely unknown. The MPV17 protein is located in the inner membrane of cell structures called mitochondria. Mitochondria are involved in a wide variety of cellular activities, including energy production, chemical signaling, and regulation of cell growth and division. Mitochondria contain their own DNA, known as mitochondrial DNA (mtDNA), which is essential for the normal function of these structures. It is likely that the MPV17 protein is involved in the maintenance of mtDNA. Having an adequate amount of mtDNA is essential for normal energy production within cells.

Health Conditions Related to Genetic Changes

MPV17-related hepatocerebral mitochondrial DNA depletion syndrome

More than 30 mutations in the *MPV17* gene have been found to cause *MPV17*-related hepatocerebral mitochondrial DNA depletion syndrome, a condition characterized by liver disease and neurological problems that begin in infancy. Most of the mutations that cause this condition change single protein building blocks (amino acids) in the MPV17 protein. One mutation that almost exclusively affects the Navajo population of the southwestern United States replaces the amino acid arginine with the amino acid glutamine at position 50 in the protein (written as R50Q). This mutation results in the production of an unstable MPV17 protein that is quickly broken down. When the condition occurs in people of Navajo ancestry, it is called Navajo neurohepatopathy.

The changes in the MPV17 protein that cause *MPV17*-related hepatocerebral mitochondrial DNA depletion syndrome, including the R50Q mutation, impair protein function and reduce the amount of protein that is available. A dysfunctional or absent MPV17 protein leads to problems with the maintenance of mtDNA, which can cause a reduction in the amount of mtDNA (known as mitochondrial DNA depletion). Mitochondrial DNA depletion impairs mitochondrial function in many of the body's cells and tissues, particularly the brain, liver, and other tissues that have high energy requirements. Reduced mitochondrial function in the liver and brain lead to the liver failure and neurological dysfunction associated with *MPV17*-related hepatocerebral mitochondrial DNA depletion syndrome. Researchers suggest that the less mtDNA that is available in cells, the more severe the features of Navajo neurohepatopathy.

Charcot-Marie-Tooth disease

MedlinePlus Genetics provides information about Charcot-Marie-Tooth disease

Other Names for This Gene

- MpV17 mitochondrial inner membrane protein
- MPV17, mitochondrial inner membrane protein
- MPV17_HUMAN
- MTDPS6
- SYM1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MPV17 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4358[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MPV17%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MITOCHONDRIAL INNER MEMBRANE PROTEIN MPV17; MPV17 (https://omim.org/entry/137960)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4358)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MPV17[gene])

References

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Genomic Location

The *MPV17* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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