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
Chromosomal Location

Cytogenetic Location: 2p23.3, which is the short (p) arm of chromosome 2 at position 23.3

Molecular Location: base pairs 27,309,492 to 27,323,097 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Other Names for This Gene

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

Additional Information & Resources

Educational Resources

  https://www.ncbi.nlm.nih.gov/books/NBK22005/#A3434
  https://www.ncbi.nlm.nih.gov/books/NBK9896/

Clinical Information from GeneReviews

- Mitochondrial DNA Maintenance Defects Overview
  https://www.ncbi.nlm.nih.gov/books/NBK487393
- MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect
  https://www.ncbi.nlm.nih.gov/books/NBK92947
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  https://www.ncbi.nlm.nih.gov/pubmed?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
• MPV17, MOUSE, HOMOLOG OF
  http://omim.org/entry/137960

Research Resources
• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MPV17%5Bgene%5D
• HGNC Gene Symbol Report
• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:4358
• NCBI Gene
• UniProt
  https://www.uniprot.org/uniprot/P39210

Sources for This Summary
• OMIM: MPV17, MOUSE, HOMOLOG OF
  http://omim.org/entry/137960
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16582910

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17694548

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