MCOLN1 gene

mucolipin 1

Normal Function

The *MCOLN1* gene provides instructions for making a protein called mucolipin-1. This protein is located in the membranes of lysosomes and endosomes, compartments within the cell that digest and recycle materials. While its function is not completely understood, mucolipin-1 plays a role in the transport (trafficking) of fats (lipids) and proteins between lysosomes and endosomes.

Mucolipin-1 acts as a channel, allowing positively charged atoms (cations) to cross the membranes of lysosomes and endosomes. It remains unclear which cations are allowed to flow through this channel. Mucolipin-1 appears to be important for the development and maintenance of the brain and light-sensitive tissue at the back of the eye (retina). In addition, this protein is likely critical for normal functioning of the cells in the stomach that produce digestive acids.

Health Conditions Related to Genetic Changes

Mucolipidosis type IV

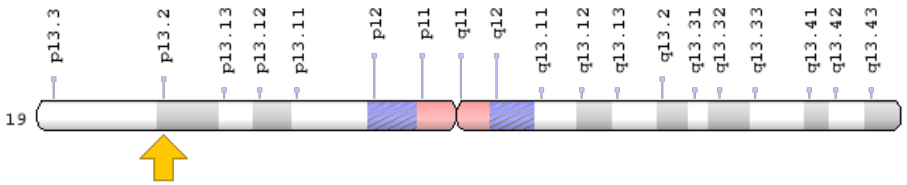
At least 22 mutations in the *MCOLN1* gene have been found to cause mucolipidosis type IV. Most of these mutations result in the production of a nonfunctional protein or prevent any protein from being produced. Two mutations in the *MCOLN1* gene account for almost all cases of mucolipidosis type IV in people with Ashkenazi Jewish ancestry. The most common mutation, written as 406-2A>G, changes a single DNA building block (nucleotide) in a region of the gene known as intron 3. This mutation, which is called a splice-site mutation, introduces a premature stop signal in the instructions for making mucolipin-1. The other mutation, written as 511_6943del, deletes a large amount of DNA near the beginning of the *MCOLN1* gene. Both of these mutations result in the production of an abnormally short, nonfunctional protein.

A lack of functional mucolipin-1 impairs transport of lipids and proteins, causing these substances to build up inside lysosomes. It remains unclear how mutations in the *MCOLN1* gene lead to delayed development of mental and motor skills (psychomotor delay), progressive vision loss, and impaired secretion of stomach acid (achlorhydia) in people with mucolipidosis type IV.

Chromosomal Location

Cytogenetic Location: 19p13.2, which is the short (p) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 7,522,610 to 7,534,009 on chromosome 19 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MCLN1_HUMAN
- ML4
- MLIV
- MST080
- MSTP080
- mucolipidin
- TRP-ML1
- TRPML1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The endocytic pathway from the plasma membrane to lysosomes
  https://www.ncbi.nlm.nih.gov/books/NBK28297/figure/A3194/

Clinical Information from GeneReviews

- Mucolipidosis IV
  https://www.ncbi.nlm.nih.gov/books/NBK1214

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MCOLN1%5BTIAB%5D%29+OR+%28mucolipin+1%5BTIAB%5D%29%29+OR+%28%28ML4%5BTIAB%5D%29+OR+%28MLIV%5BTIAB%5D%29+OR+%28TRPML1%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+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- MUCOLIPIN 1
  http://omim.org/entry/605248

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MCOLN1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MCOLN1%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:13356
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:57192
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/57192
- UniProt
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