

## MCOLN1 gene

mucolipin TRP cation channel 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

#### Mucopolidosis type IV

At least 22 mutations in the *MCOLN1* gene have been found to cause mucopolidosis 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 mucopolidosis 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 (achlorhydria) in people with mucopolidosis type IV.

## Other Names for This Gene

- MCLN1\_HUMAN
- ML4
- MLIV
- MST080
- MSTP080
- mucolipidin
- TRP-ML1
- TRPML1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of MCOLN1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57192\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57192[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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; MCOLN1 (<https://omim.org/entry/605248>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/57192>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=MCOLN1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=MCOLN1[gene]))

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## Genomic Location

The *MCOLN1* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chro>)

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