

NPC1 gene

NPC intracellular cholesterol transporter 1

Normal Function

The *NPC1* gene provides instructions for making a protein that is located within the membrane of compartments in the cell called lysosomes and endosomes, which digest and recycle materials. While the exact function of this protein is unclear, it plays a role in the movement of cholesterol and other types of fats (lipids) within cells and across cell membranes.

Health Conditions Related to Genetic Changes

Niemann-Pick disease

More than 380 mutations in the *NPC1* gene have been found to cause Niemann-Pick disease type C1. This type of Niemann-Pick disease is characterized by a buildup of fat within cells that leads to movement problems, neurological impairment, lung and liver disease, and speech and feeding problems. Many of the *NPC1* gene mutations result in a change in a single protein building block (amino acid) in the NPC1 protein. These mutations usually cause a shortage of functional protein, which prevents movement of cholesterol and other lipids, leading to their accumulation in cells. Because these lipids are not in their proper location in cells, many normal cell functions that require lipids (such as cell membrane formation) are impaired. The accumulation of lipids and the cell dysfunction eventually leads to cell death, causing the tissue and organ damage seen in Niemann-Pick disease type C1.

Other Names for This Gene

- Niemann-Pick disease, type C1
- NPC

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NPC1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4864\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4864[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28NPC1%5BTI%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- NPC INTRACELLULAR CHOLESTEROL TRANSPORTER 1; NPC1 (<https://omim.org/entry/607623>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4864>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NPC1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NPC1[gene]))

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Genomic Location

The *NPC1* gene is found on chromosome 18 (<https://medlineplus.gov/genetics/chromosome/18/>).

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