

## **NPC2 gene**

NPC intracellular cholesterol transporter 2

### **Normal Function**

The *NPC2* gene provides instructions for making a protein that is located inside lysosomes, which are compartments in the cell that digest and recycle materials. The NPC2 protein attaches (binds) to cholesterol. Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods. Research suggests that the NPC2 protein plays an important role in moving cholesterol and certain other fats (lipids) out of the lysosomes to other parts of the cell.

### **Health Conditions Related to Genetic Changes**

#### Niemann-Pick disease

More than 20 mutations in the *NPC2* gene have been found to cause Niemann-Pick disease type C2. 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. The *NPC2* gene mutations that cause Niemann-Pick disease type C2 reduce or eliminate NPC2 protein activity, 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 C2.

### **Other Names for This Gene**

- epididymal secretory protein
- HE1
- MGC1333
- Niemann-Pick disease, type C2
- NP-C2
- NPC2\_HUMAN

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of NPC2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10577\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10577[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28NPC2%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+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- NPC INTRACELLULAR CHOLESTEROL TRANSPORTER 2; NPC2 (<https://omim.org/entry/601015>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10577>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NPC2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NPC2[gene]))

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## Genomic Location

The *NPC2* gene is found on chromosome 14 (<https://medlineplus.gov/genetics/chromosome/14/>).

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