

## NEU1 gene

neuraminidase 1

### Normal Function

The *NEU1* gene provides instructions for making an enzyme called neuraminidase 1 (NEU1), which is found in lysosomes. Lysosomes are compartments within cells that use enzymes to digest and recycle materials. The NEU1 enzyme helps break down large sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins) by removing an substance known as sialic acid.

### Health Conditions Related to Genetic Changes

#### Sialidosis

At least 42 mutations in the *NEU1* gene have been found to cause sialidosis. Most of these mutations change single protein building blocks (amino acids) used to make the NEU1 enzyme. Mutations in the *NEU1* gene lead to a shortage (deficiency) of the NEU1 enzyme. When this enzyme is lacking, large molecules that are usually broken down by the NEU1 enzyme accumulate inside lysosomes. Conditions such as sialidosis that cause large molecules to build up inside lysosomes are called lysosomal storage disorders. Mutations that eliminate NEU1 enzyme activity cause more severe signs and symptoms than those that result in some functional enzyme. It is unclear exactly how the accumulation of large molecules within lysosomes leads to the signs and symptoms of sialidosis.

### Other Names for This Gene

- acetylneuraminyl hydrolase
- exo-alpha-sialidase
- FLJ93471
- G9 sialidase
- lysosomal sialidase
- N-acetyl-alpha-neuraminidase 1
- NANH
- NEU
- NEUR1\_HUMAN

- neuraminidase 1 (lysosomal sialidase)
- neuraminidase 1 precursor
- SIAL1
- sialidase 1
- sialidase 1 (lysosomal sialidase)

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of NEU1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4758\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4758[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28NEU1%5BTIAB%5D%29+OR+%28%28neuraminidase+1%5BTIAB%5D%29+OR+%28lysosomal+sialidase%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+720+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- NEURAMINIDASE 1; NEU1 (<https://omim.org/entry/608272>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4758>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=NEU1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=NEU1[gene]))

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## Genomic Location

The *NEU1* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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