

## HEXA gene

hexosaminidase subunit alpha

### Normal Function

The *HEXA* gene provides instructions for making one part (subunit) of an enzyme called beta-hexosaminidase A. Specifically, the protein produced from the *HEXA* gene forms the alpha subunit of this enzyme. One alpha subunit joins with one beta subunit (produced from the *HEXB* gene) to form a functioning beta-hexosaminidase A enzyme.

Beta-hexosaminidase A plays a critical role in the brain and spinal cord (central nervous system). This enzyme is found in lysosomes, which are structures in cells that break down toxic substances and act as recycling centers. Within lysosomes, beta-hexosaminidase A forms part of a complex that breaks down a fatty substance called GM2 ganglioside found in cell membranes.

### Health Conditions Related to Genetic Changes

#### Tay-Sachs disease

More than 210 variants (also known as mutations) that cause Tay-Sachs disease have been identified in the *HEXA* gene. Tay-Sachs disease is a condition that is characterized by movement disorders, intellectual and developmental disability, and other neurological problems caused by the death of nerve cells (neurons) in the central nervous system.

The *HEXA* gene variants that cause Tay-Sachs disease eliminate or severely reduce the activity of the enzyme beta-hexosaminidase A. This lack of enzyme activity prevents the enzyme from breaking down GM2 ganglioside. As a result, this substance builds up to toxic levels, particularly in neurons in the central nervous system. Progressive damage caused by the buildup of GM2 ganglioside leads to the destruction of these cells, which causes the signs and symptoms of Tay-Sachs disease.

Most of the known *HEXA* gene variants result in a completely nonfunctional version of beta-hexosaminidase A. These variants cause a severe form of Tay-Sachs disease, known as infantile Tay-Sachs disease, which appears in infancy. Other variants severely reduce but do not eliminate the activity of beta-hexosaminidase A. These genetic changes are responsible for less severe forms of Tay-Sachs disease, known as the juvenile and late-onset forms, which appear later in life.

## Other Names for This Gene

- Beta-hexosaminidase A
- beta-N-Acetylhexosaminidase A
- Hex A
- HEXA\_HUMAN
- hexosaminidase A (alpha polypeptide)
- N-acetyl-beta-glucosaminidase

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of HEXA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3073\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3073[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28HEXA+AND+hexosaminidase%5BTIAB%5D%29+OR+%28hexosaminidase+A%5BTIAB%5D%29%29+OR+%28%28Beta-hexosaminidase+A%5BTIAB%5D%29+OR+%28beta-N-Acetylhexosaminidase+A%5BTIAB%5D%29+OR+%28N-acetyl-beta-glucosaminidase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- HEXOSAMINIDASE A; HEXA (<https://omim.org/entry/606869>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3073>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=HEXA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=HEXA[gene]))

## References

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## Genomic Location

The *HEXA* gene is found on chromosome 15 (<https://medlineplus.gov/genetics/chromosome/15/>).

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