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 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.

Health Conditions Related to Genetic Changes

Tay-Sachs disease

More than 120 mutations that cause Tay-Sachs disease have been identified in the $HEXA$ gene. These mutations reduce or eliminate the activity of the enzyme beta-hexosaminidase A, which prevents the enzyme from breaking down GM2 ganglioside. As a result, this substance builds up to toxic levels, particularly in nerve cells in the brain and spinal cord. 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 $HEXA$ mutations result in a completely nonfunctional version of beta-hexosaminidase A. These mutations cause the severe form of Tay-Sachs disease, which appears in infancy. Other mutations reduce but do not eliminate the activity of beta-hexosaminidase A; these genetic changes are responsible for the less severe forms of Tay-Sachs disease, which appear later in life.
Chromosomal Location

Cytogenetic Location: 15q23, which is the long (q) arm of chromosome 15 at position 23

Molecular Location: base pairs 72,343,435 to 72,376,473 on chromosome 15 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

Credit: Genome Decoration Page/NCBI

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

Clinical Information from GeneReviews

• Hexosaminidase A Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1218

Scientific Articles on PubMed

• PubMed
Catalog of Genes and Diseases from OMIM

- HEXOSAMINIDASE A
  http://omim.org/entry/606869

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_HEXNA.html

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=HEXA%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3073

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P06865

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9572057

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301397

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16698036
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2910082/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10571007

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14577003
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16088929

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12019216

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