HEXB gene
hexosaminidase subunit beta

Normal Function

The HEXB gene provides instructions for making a protein that is a part (subunit) of two related enzymes, beta-hexosaminidase A and beta-hexosaminidase B. Each of these enzymes is made up of two subunits. Beta-hexosaminidase A includes one alpha subunit (produced from the HEXA gene) and one beta subunit (produced from the HEXB gene). Beta-hexosaminidase B is composed of two beta subunits, which are produced from the HEXB gene.

Beta-hexosaminidase A and beta-hexosaminidase B play a critical role in the brain and spinal cord (central nervous system). These enzymes are found in lysosomes, which are structures in cells that break down toxic substances and act as recycling centers. Within lysosomes, the enzymes break down fatty compounds called sphingolipids, complex sugars called oligosaccharides, and molecules that are linked to sugars (such as glycoproteins). In particular, beta-hexosaminidase A forms part of a complex that breaks down a fatty substance called GM2 ganglioside.

Health Conditions Related to Genetic Changes

Sandhoff disease

About 30 mutations that cause Sandhoff disease have been identified in the HEXB gene. These mutations reduce or eliminate the activity of both beta-hexosaminidase A and beta-hexosaminidase B. The malfunctioning or missing enzymes are unable to break down GM2 ganglioside and other molecules, which allows these compounds to accumulate within cells. Increased levels of GM2 ganglioside are particularly toxic to nerve cells in the central nervous system. Excess GM2 ganglioside leads to the progressive destruction of these cells, which causes many of the characteristic features of Sandhoff disease.

Most of the known mutations in the HEXB gene cause the severe form of Sandhoff disease, which becomes apparent in infancy. These mutations prevent cells from making any beta-hexosaminidase A or beta-hexosaminidase B, or lead to the production of completely nonfunctional versions of these enzymes. The most common mutation deletes a large segment of DNA near the beginning of the HEXB gene, which results in a total loss of enzyme activity. Other mutations reduce but do not eliminate the activity of the enzymes; these genetic changes are responsible for the less severe forms of Sandhoff disease, which appear later in life.
Chromosomal Location

Cytogenetic Location: 5q13.3, which is the long (q) arm of chromosome 5 at position 13.3

Molecular Location: base pairs 74,640,023 to 74,721,288 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

• beta-N-acetylhexosaminidase B
• ENC-1AS
• Hex B
• HEXB_HUMAN
• hexosaminidase B
• hexosaminidase B (beta polypeptide)

Additional Information & Resources

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HEXB%5BTIAB%5D%29+OR+%28hexosaminidase+B%5BTIAB%5D%29+OR+%28ENC-1AS%5BTIAB%5D%29+OR+%28hexosaminidase+B%5BTIAB%5D%29+OR+%28N-acetyl-beta-glucosaminidase%5BTIAB%5D%29+OR+%28Hex+B%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D+OR+Genetic+Phenomena%5BMH%5D+OR+human%5Bmh%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND%22last+2160+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

• HEXOSAMINIDASE B
  http://omim.org/entry/606873
Research Resources

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

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

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:3074

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P07686

Sources for This Summary


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