GM2A gene
GM2 ganglioside activator

Normal Function

The GM2A gene provides instructions for making a protein called the GM2 ganglioside activator. This protein is necessary for the normal function of an enzyme called beta-hexosaminidase A, which plays a critical role in the brain and spinal cord (central nervous system). Beta-hexosaminidase A and the GM2 ganglioside activator protein work together in lysosomes, which are structures in cells that break down toxic substances and act as recycling centers. Within lysosomes, the activator protein binds to a fatty substance called GM2 ganglioside and presents it to beta-hexosaminidase A to be broken down.

Health Conditions Related to Genetic Changes

GM2-gangliosidosis, AB variant

Only a few mutations in the GM2A gene have been identified in people with GM2-gangliosidosis, AB variant. Some of these mutations change single protein building blocks (amino acids) in the GM2 ganglioside activator. Other mutations delete a small amount of DNA from the GM2A gene. These genetic changes result in an unstable activator protein that is quickly degraded, or they prevent the gene from making any functional protein. Without the GM2 ganglioside activator, beta-hexosaminidase A is unable to break 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 the AB variant.
Chromosomal Location

Cytogenetic Location: 5q33.1, which is the long (q) arm of chromosome 5 at position 33.1

Molecular Location: base pairs 151,253,185 to 151,270,440 on chromosome 5 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- cerebroside sulfate activator protein
- GM2 activator
- GM2 ganglioside activator protein
- SAP-3
- SAP3_HUMAN
- sphingolipid activator protein 3

Additional Information & Resources

Educational Resources
  [Link](https://www.ncbi.nlm.nih.gov/books/NBK20729/#A1387)

Scientific Articles on PubMed
- PubMed
  [Link](https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GM2A%5BTIAB%5D%29+OR+%28GM2+ganglioside+activator%5BTIAB%5D%29%29+OR+%28GM2+activator%5BTIAB%5D%29+OR+%28SAP-3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)
Catalog of Genes and Diseases from OMIM

- GM2 ACTIVATOR
  http://omim.org/entry/613109

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_GM2A.html

- ClinVar

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:2760

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/P17900

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10364519
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1378077/

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

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9714704

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8900233
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