GM2A gene

ganglioside GM2 activator

Normal Function

The *GM2A* gene provides instructions for making a protein called the ganglioside GM2 activator. This protein is necessary for the normal function of an enzyme called beta-hexosaminidase A. Beta-hexosaminidase A and the ganglioside GM2 activator protein work together in lysosomes, which are compartments in the cell that digest and recycle different types of molecules. 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 activator deficiency

A few variants (also called mutations) in the *GM2A* gene have been identified in people with GM2 activator deficiency (sometimes called GM2 gangliosidosis, AB variant). This is a rare inherited disorder that results in progressive brain injury. Some of the *GM2A* gene variants change single protein building blocks (amino acids) in the ganglioside GM2 activator. Other variants delete a small amount of DNA from the *GM2A* gene. Some of these genetic changes result in an unstable activator protein that quickly breaks down, while others prevent the gene from making any functional protein. Without the ganglioside GM2 activator, beta-hexosaminidase A is unable to break down GM2 ganglioside. As a result, this substance builds up to toxic levels. Progressive damage caused by the buildup of GM2 ganglioside leads to the typical signs and symptoms of GM2 activator deficiency.

Other Names for This Gene

- ganglioside GM2 activator
- GM2 activator
- GM2 ganglioside activator
- GM2-AP
- GM2AP
- SAP-3

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GM2A (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2760[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GM2A%5BTIAB%5D%29+OR+%28GM2+ganglioside+activator%5BTIAB%5D%29%29+OR+%28%28cerebroside+sulfate+activator+protein%5BTIAB%5D%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%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; GM2A (https://omim.org/entry/613109)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2760)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GM2A[gene])

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Genomic Location

The *GM2A* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

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