SMPD1 gene

sphingomyelin phosphodiesterase 1

Normal Function

The *SMPD1* gene provides instructions for making an enzyme called acid sphingomyelinase. This enzyme is found in lysosomes, which are small compartments in the cell that digest and recycle molecules. Acid sphingomyelinase is responsible for the conversion of a fat (lipid) called sphingomyelin into another type of lipid called ceramide. Sphingomyelin also binds (attaches) to a fat called cholesterol and helps to form other lipids that play roles in various cell processes. The formations of these lipids is critical for the normal structure and function of cells and tissues.

Health Conditions Related to Genetic Changes

Niemann-Pick disease

At least 175 mutations in the *SMPD1* gene have been found to cause Niemann-Pick disease types A and B. These types of Niemann-Pick disease are characterized by a buildup of fat within cells that leads to lung disease and enlargement of the liver and spleen (hepatosplenomegaly). Type A is more severe and is characterized by severe neurological impairment in early childhood.

SMPD1 gene mutations that cause complete loss of enzyme function tend to cause Niemann-Pick disease type A. In the Ashkenazi (eastern and central European) Jewish population, three mutations are responsible for about 90 percent of all Niemann-Pick disease type A cases. Mutations that lead to the production of an enzyme that retains some activity often cause Niemann-Pick disease type B. A reduction in enzyme activity within cells allows sphingomyelin to accumulate in cells. The accumulation of this lipid causes cells to malfunction and eventually die. Over time, cell loss impairs function of tissues and organs including the brain, lungs, spleen, and liver in people with Niemann-Pick disease types A and B.

Other Names for This Gene

- acid sphingomyelinase
- ASM
- ASM_HUMAN

- sphingomyelin phosphodiesterase 1, acid lysosomal
- sphingomyelin phosphodiesterase 1, acid lysosomal (acid sphingomyelinase)

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SMPD1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6609[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SMPD1%5BTIAB%5D%29+OR+%28sphingomyelin+phosphodiesterase+1%5BALL%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

- SPHINGOMYELIN PHOSPHODIESTERASE 1, ACID LYSOSOMAL; SMPD1 (https://omim.org/entry/607608)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6609)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SMPD1[gene])

References

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Genomic Location

The *SMPD1* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

Last updated January 1, 2015