SGSH gene

N-sulfoglucosamine sulfohydrolase

Normal Function

The *SGSH* gene provides instructions for producing an enzyme called sulfamidase. This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. Sulfamidase is involved in the step-wise breakdown of large molecules called glycosaminoglycans (GAGs). GAGs are composed of sugar molecules that are linked together to form a long string. To break down these large molecules, individual sugars are removed one at a time from one end of the molecule. Sulfamidase removes a chemical group known as a sulfate from a sugar called glucosamine when it is at the end of the GAG chain.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type III

More than 80 mutations in the *SGSH* gene have been found to cause mucopolysaccharidosis type IIIA (MPS IIIA). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IIIA reduce or eliminate the function of sulfamidase.

The lack of sulfamidase activity disrupts the breakdown of a subset of GAGs called heparan sulfate. As a result, partially broken down heparan sulfate accumulates within lysosomes. Researchers believe that the accumulation of GAGs interferes with the functions of other proteins inside the lysosomes and disrupts the normal functions of cells. It is unknown why the buildup of heparan sulfate mostly affects the central nervous system in MPS IIIA.

Other Names for This Gene

- heparan N-sulfatase
- heparan sulfate sulfatase
- HSS
- N-sulphoglucosamine sulphohydrolase
- N-sulphoglucosamine sulphohydrolase precursor
- SFMD

- SPHM_HUMAN
- sulfamidase
- sulfoglucosamine sulfamidase
- sulphamidase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SGSH (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6448[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SGSH%5BTIAB%5D%29+OR+%28N-sulfoglucosamine+sulfohydrolase%5BTIAB%5D%29%29+OR+%28%28heparan+sulfate+sulfatase%5BTIAB%5D%29+OR+%28MPS3A%5BTIAB%5D%29+OR+%28N-sulphoglucosamine+sulphohydrolase%5BTIAB%5D%29+OR+%28sulphamidase%5BTIAB%5D%29+OR+%28heparan+N-sulfatase%5BTIAB%5D%29+OR+%28sulfamidase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- N-SULFOGLUCOSAMINE SULFOHYDROLASE; SGSH (https://omim.org/entry/605270)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6448)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SGSH[gene])

References

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- Muschol N, Storch S, Ballhausen D, Beesley C, Westermann JC, Gal A, Ullrich K, Hopwood JJ, Winchester B, Braulke T. Transport, enzymatic activity, and stabilityof mutant sulfamidase (SGSH) identified in patients with mucopolysaccharidosistype III

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- Valstar MJ, Ruijter GJ, van Diggelen OP, Poorthuis BJ, Wijburg FA. Sanfilipposyndrome: a mini-review. J Inherit Metab Dis. 2008 Apr;31(2):240-52. doi: 10.1007/s10545-008-0838-5. Epub 2008 Apr 4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18392742)

Genomic Location

The *SGSH* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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