

GNS gene

glucosamine (N-acetyl)-6-sulfatase

Normal Function

The *GNS* gene provides instructions for producing an enzyme called N-acetylglucosamine-6-sulfatase. This enzyme is located in lysosomes, compartments within cells that digest and recycle different types of molecules. N-acetylglucosamine-6-sulfatase 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. N-acetylglucosamine-6-sulfatase removes a chemical group known as a sulfate from a subset of GAGs called heparan sulfate when the sugar N-acetylglucosamine-6-sulfate is located at the end.

Health Conditions Related to Genetic Changes

Mucopolysaccharidosis type III

Mutations in the *GNS* gene cause mucopolysaccharidosis type IIID (MPS IIID). Most of these mutations change single DNA building blocks (nucleotides) in the gene. All of the mutations that cause MPS IIID reduce or eliminate the function of N-acetylglucosamine-6-sulfatase.

The lack of N-acetylglucosamine-6-sulfatase activity disrupts the breakdown of heparan sulfate. As a result, partially broken down GAGs accumulate 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 IIID.

Other Names for This Gene

- G6S
- glucosamine-6-sulfatase
- GNS_HUMAN
- MGC21274
- N-acetylglucosamine-6-sulfatase

- N-acetylglucosamine-6-sulfatase precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GNS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2799\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2799[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GNS%5BTIAB%5D%29+OR+%28glucosamine-6-sulfatase%5BTIAB%5D%29+AND+%28mucopolysaccharidosis%5BTIAB%5D%29%29+OR+%28%28N-acetylglucosamine-6-sulfatase%5BTIAB%5D%29+OR+%28glucosamine-6-sulfatase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+NOT+%28%28Hedgehog%29+OR+%28coevolution%5BTI%5D%29%29+AND+englis h%5Bla%5D%29>)

Catalog of Genes and Diseases from OMIM

- N-ACETYLGLUCOSAMINE-6-SULFATASE; GNS (<https://omim.org/entry/607664>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/2799>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GNS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GNS[gene]))

References

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- Valstar MJ, Ruijter GJ, van Diggelen OP, Poorthuis BJ, Wijburg FA. Sanfilipposyndrometype D: a mini-review. J Inher 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 *GNS* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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