NAGA gene

alpha-N-acetylgalactosaminidase

Normal Function

The *NAGA* gene provides instructions for making the enzyme alpha-N-acetylgalactosaminidase. This enzyme works in the lysosomes, which are compartments within cells that digest and recycle materials. Within lysosomes, the enzyme helps break down complexes called glycoproteins and glycolipids, which consist of sugar molecules attached to certain proteins and fats. Specifically, alpha-N-acetylgalactosaminidase helps remove a molecule called alpha-N-acetylgalactosamine from sugars in these complexes.

Health Conditions Related to Genetic Changes

Schindler disease

Approximately seven *NAGA* gene mutations have been identified in people with Schindler disease. Most of these mutations are believed to change the 3-dimensional shape of the alpha-N-acetylgalactosaminidase enzyme, interfering with its ability to break down glycoproteins and glycolipids. These substances accumulate in the lysosomes and cause cells to malfunction and eventually die. Cell damage in the various tissues and organs of the body leads to neurological problems and the other signs and symptoms of Schindler disease.

Other Names for This Gene

- Acetylgalactosaminidase, alpha-N- (alpha-galactosidase B)
- alpha-N-acetylgalactosaminidase precursor
- D22S674
- GALB
- N-acetylgalactosaminidase, alpha-
- NAGAB_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of NAGA (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4668[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28NAGA%5BTIAB%5D%29+OR+%28%28alpha-N-acetylgalactosaminidase%5BTIAB%5D%29+OR+%28alpha-N-acetylgalactosaminidase+precursor%5BTIAB%5D%29+OR+%28GALB%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- N-ACETYL-ALPHA-D-GALACTOSAMINIDASE; NAGA (https://omim.org/entry/104170)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4668)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=NAGA[gene])

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Genomic Location

The *NAGA* gene is found on chromosome 22 (https://medlineplus.gov/genetics/chromosome/22/).

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