

## AGA gene

aspartylglucosaminidase

### Normal Function

The *AGA* gene provides instructions for producing an enzyme called aspartylglucosaminidase. This enzyme is active in lysosomes, which are structures inside cells that act as recycling centers. Within lysosomes, the enzyme helps break down complex chains of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins). Specifically, this enzyme cuts (cleaves) glycoproteins between a protein building block (amino acid) called asparagine and a sugar molecule called N-acetylglucosamine. This cut is one of the last steps in breaking down a glycoprotein in the lysosome.

### Health Conditions Related to Genetic Changes

#### Aspartylglucosaminuria

Many variants (also known as mutations) in the *AGA* gene have been found to cause aspartylglucosaminuria. This condition primarily affects mental functioning and movement. Aspartylglucosaminuria worsens over time. Most *AGA* gene variants change one amino acid in aspartylglucosaminidase. One variant, found in 98 percent of people with this condition in Finland, replaces the amino acid cysteine with the amino acid serine at position 163 in the enzyme (written as Cys163Ser or C163S).

Many *AGA* gene variants, including C163S, disrupt the structure of aspartylglucosaminidase, resulting in an enzyme that cannot effectively break down glycoproteins because it cannot make the cut between asparagine and N-acetylglucosamine. A buildup of glycoproteins seems to particularly affect nerve cells in the brain; loss of these cells causes a progressive decline in mental functioning and the other signs and symptoms of aspartylglucosaminuria.

### Other Names for This Gene

- ASRG
- glycosylasparaginase
- N(4)-(beta-N-acetylglucosaminy)-L-asparaginase
- N4-(N-acetyl-beta-glucosaminy)-L-asparagine amidase

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of AGA ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=175\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=175[geneid]))

### Scientific Articles on PubMed

- PubMed ([https://pubmed.ncbi.nlm.nih.gov/?term=\(\(aspartylglucosaminidase%5BTIAB%5D\)+OR+\(glycosylasparaginase%5BTIAB%5D\)\)+AND+english%5Bla%5D+AND+human%5Bmh%5D\)](https://pubmed.ncbi.nlm.nih.gov/?term=((aspartylglucosaminidase%5BTIAB%5D)+OR+(glycosylasparaginase%5BTIAB%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D)))

### Catalog of Genes and Diseases from OMIM

- ASPARTYLGLUCOSAMINIDASE; AGA (<https://omim.org/entry/613228>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/175>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=AGA\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AGA[gene]))

## References

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## **Genomic Location**

The *AGA* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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