

PSEN2 gene

presenilin 2

Normal Function

The *PSEN2* gene provides instructions for making a protein called presenilin 2. Presenilin 2 helps process proteins that transmit chemical signals from the cell membrane into the nucleus. Once in the nucleus, these signals turn on (activate) genes that are important for cell growth and maturation.

Presenilin 2 is best known for its role in processing amyloid precursor protein, which is found in the brain and other tissues. Research suggests that presenilin 2 works together with other enzymes to cut amyloid precursor protein into smaller segments (peptides). One of these peptides is called soluble amyloid precursor protein (sAPP), and another is called amyloid beta peptide. Recent evidence suggests that sAPP has growth-promoting properties and may play a role in the formation of neurons in the brain both before and after birth. Other functions of sAPP and amyloid beta peptide are under investigation.

Health Conditions Related to Genetic Changes

Alzheimer's disease

At least 11 mutations in the *PSEN2* gene have been shown to cause early-onset Alzheimer's disease. Mutations in this gene account for less than 5 percent of all early-onset cases of the disorder.

Two of the most common *PSEN2* mutations that cause early-onset Alzheimer's disease change single protein building blocks (amino acids) used to make presenilin 2. One mutation replaces the amino acid asparagine with the amino acid isoleucine at position 141 (written as Asn141Ile or N141I). The other mutation changes the amino acid methionine to the amino acid valine at position 239 (written as Met239Val or M239V). These mutations appear to disrupt the processing of amyloid precursor protein, leading to the overproduction of amyloid beta peptide. This protein fragment can build up in the brain and form clumps called amyloid plaques that are characteristic of Alzheimer's disease. A buildup of toxic amyloid beta peptide and amyloid plaques may lead to the death of neurons and the progressive signs and symptoms of this disorder.

Familial dilated cardiomyopathy

MedlinePlus Genetics provides information about Familial dilated cardiomyopathy

Other Names for This Gene

- AD3-like protein
- AD3L
- AD3LP
- AD4
- AD5
- Alzheimer's disease 3-like
- E5-1
- presenilin 2 (Alzheimer disease 4)
- PS2 protein (alzheimer-associated)
- PSN2_HUMAN
- PSNL2
- STM2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PSEN2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5664\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5664[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PSEN2%5BTIAB%5D%29+OR+%28presenilin+2%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+1080+days%22%5Bdp%5D%29%29%29>)

Catalog of Genes and Diseases from OMIM

- PRESENILIN 2; PSEN2 (<https://omim.org/entry/600759>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5664>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PSEN2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PSEN2[gene]))

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Genomic Location

The *PSEN2* gene is found on chromosome 1 (<https://medlineplus.gov/genetics/chromosome/1/>).

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