CST3 gene

cystatin C

Normal Function

The *CST3* gene provides instructions for making a protein called cystatin C. This protein is part of a family of proteins called cysteine protease inhibitors that help control several types of chemical reactions by blocking (inhibiting) the activity of certain enzymes. Cystatin C inhibits the activity of enzymes called cathepsins that cut apart other proteins in order to break them down.

Cystatin C is found in biological fluids, such as blood. Its levels are especially high in the fluid that surrounds and protects the brain and spinal cord (the cerebrospinal fluid or CSF).

Health Conditions Related to Genetic Changes

Hereditary cerebral amyloid angiopathy

At least one variant (also called a mutation) in the *CST3* gene has been found to cause hereditary cerebral amyloid angiopathy, a condition characterized by stroke and a decline in intellectual function (dementia), which begins in mid-adulthood. The *CST3* gene variant that has been identified causes a form of the condition known as hereditary cerebral hemorrhage, Icelandic type or Icelandic type cerebral amyloid angiopathy. This variant replaces the protein building block (amino acid) leucine with the amino acid glutamine at position 68 in the cystatin C protein (written as Leu68Gln or L68Q). This abnormal cystatin C protein is less stable and is more prone to cluster together (aggregate) than the normal protein. The aggregated protein forms clumps called amyloid deposits that accumulate in the blood vessel walls primarily in the brain, but also in blood vessels in other areas of the body such as the skin, spleen, and lymph nodes. The accumulation of these amyloid deposits, does not appear to have any health effects outside of the brain. In the brain, the amyloid deposits replace the muscle fibers and elastic fibers that give blood vessels flexibility, causing them to become weak and prone to breakage. Such a break in the brain causes bleeding (hemorrhagic stroke), which can lead to brain damage and dementia.

Age-related macular degeneration

MedlinePlus Genetics provides information about Age-related macular degeneration

Other Names for This Gene

- cystatin 3
- cystatin-3
- cystatin-C
- cystatin-C precursor
- CYTC_HUMAN
- gamma-trace
- neuroendocrine basic polypeptide
- post-gamma-globulin

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of CST3 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1471[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28CST3%5BTIAB%5D%29+OR+%28cystatin+C%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+720+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- CYSTATIN 3; CST3 (https://omim.org/entry/604312)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/1471)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=CST3[gene])

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Genomic Location

The *CST3* gene is found on chromosome 20 (https://medlineplus.gov/genetics/chromosome/20/).

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