SERPING1 gene

serpin family G member 1

Normal Function

The *SERPING1* gene provides instructions for making a protein called C1 inhibitor (C1-INH), which is a type of serine protease inhibitor (serpin). Serpins help control several types of chemical reactions by blocking the activity of certain proteins. C1-INH is important for controlling a range of processes involved in maintaining blood vessels, including inflammation. Inflammation is a normal body response to infection, irritation, or injury.

C1-INH blocks the activity of several proteins in the blood, including plasma kallikrein and the activated form of factor XII (called factor XIIa). These two proteins are involved in the production of bradykinin. Bradykinin is a protein fragment (peptide) that promotes inflammation by allowing fluids to leak through blood vessel walls into body tissues (vascular permeability). C1-INH attaches (binds) to plasma kallikrein and factor XIIa, which prevents them from completing any further reactions. These proteins are cleared from the bloodstream once they are bound to C1-INH.

Health Conditions Related to Genetic Changes

Hereditary angioedema

Hundreds of variants (also called mutations) in the *SERPING1* gene have been found to cause hereditary angioedema, which is a disorder characterized by recurrent episodes of severe swelling (angioedema). Variants in the *SERPING1* gene cause a form of the condition known as hereditary angioedema due to C1-INH deficiency. Hereditary angioedema due to C1-INH deficiency is further divided into two types: type I occurs when the C1-INH protein levels are low, and type II occurs when C1-INH is not functioning correctly.

The variants that cause hereditary angioedema due to C1-INH deficiency type I occur throughout the gene and lead to reduced levels of C1-INH in the blood. The variants that cause hereditary angioedema due to C1-INH deficiency type II usually occur in a specific region of the gene called exon 8. These variants lead to the production of a C1-INH that functions abnormally.

Without the proper levels of functional C1-INH, the activity of plasma kallikrein and

factor XIIa cannot be blocked, and excessive amounts of bradykinin are produced. Excess fluids leak through blood vessel walls and accumulate in body tissues, leading to the recurrent episodes of swelling seen in individuals with hereditary angioedema due to C1-INH deficiency.

Other Names for This Gene

- C1-INH
- C1IN
- C1INH
- C1NH
- complement component 1 inhibitor
- IC1_HUMAN
- plasma protease C1 inhibitor
- serine/cysteine proteinase inhibitor clade G member 1
- serpin peptidase inhibitor, clade G (C1 inhibitor), member 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SERPING1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=710[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=(SERPING1%5BTIAB%5D)+OR+((C1IN%5BTIAB%5D)+OR+(C1NH%5BTIAB%5D)+OR+(C1INH%5BTIAB%5D)+OR+(C1-INH%5BTIAB%5D))+AND+((Genes%5BMH%5D)+OR+(Genetic+Phenomena%5BMH%5D))+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- COMPLEMENT COMPONENT 1 INHIBITOR; C1NH (https://omim.org/entry/606860)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/710)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SERPING1[gene])

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Genomic Location

The *SERPING1* gene is found on chromosome 11 (https://medlineplus.gov/genetics/chromosome/11/).

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