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Variant: NM_000488.4(SERPINC1):c.1190C>G (p.Ser397Ter)

Version: 1.0

CA343772780

1068471 (ClinVar)

Gene: SERPINC1 (HGNC:462)
Condition: hereditary antithrombin deficiency (MONDO:0013144)
Inheritance Mode: Autosomal dominant inheritance
UUID: 6a951e51-a7df-493d-8016-6b5f271651a9
Approved on: 2025/10/21
Published on: 2026/1/22

HGVS expressions

NM_000488.4:c.1190C>G
NM_000488.4(SERPINC1):c.1190C>G (p.Ser397Ter)
NC_000001.11:g.173907478G>C
CM000663.2:g.173907478G>C
NC_000001.10:g.173876616G>C
CM000663.1:g.173876616G>C
NC_000001.9:g.172143239G>C
NG_012462.1:g.14901C>G
ENST00000367698.4:c.1190C>G
ENST00000367698.3:c.1190C>G
ENST00000617423.4:c.575C>G
NM_000488.3:c.1190C>G
NM_001365052.1:c.1046C>G
NM_001365052.2:c.1046C>G
NM_001386302.1:c.1313C>G
NM_001386303.1:c.1271C>G
NM_001386304.1:c.1169C>G
NM_001386305.1:c.1133C>G
NM_001386306.1:c.974C>G

Likely Pathogenic

Met criteria codes 4

PP1 | PVS1_Strong | PM2_Supporting | PS4_Supporting

Evidence Links 0

Expert Panel

Thrombosis VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen Thrombosis Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for SERPINC1 Version 1.1.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Thrombosis VCEP

The c.1190C>G; p.Ser397Ter variant in SERPINC1 is a nonsense variant that may cause loss of function of the protein, however it is predicted to escape nonsense mediated decay and remove >10% of the protein (PVS1_Strong). This variant has been reported in one proband meeting an antithrombin activity level of < 0.8 IU/mL without confirmation of repeated independent samples tested, but has a family history of antithrombin activity levels of < 0.8 IU/mL (PS4_Supporting; PMID:11307839). The variant has been reported to segregate with hereditary antithrombin deficiency in two affected family members from one family (PP1; PMID:11307839). This variant is absent from gnomAD v4.1.0 (PM2_Supporting). In summary, this variant meets the criteria to be classified as likely pathogenic for autosomal dominant hereditary antithrombin deficiency based on the ACMG/AMP criteria applied, as specified by the ClinGen Thrombosis VCEP: PVS1_Strong, PP1, PS4_Supporting, PM2_Supporting

Met criteria codes

PP1 ✔	The variant has been reported to segregate with hereditary antithrombin deficiency in two affected family members from one family (PP1; PMID:11307839).
PVS1_Strong ✔	The c.1190C>G (p.Ser397Ter) variant in SERPINC1 is a nonsense variant that may cause loss of function of the protein, however it is predicted to escape nonsense mediated decay and remove >10% of the protein (PVS1_Strong).
PM2_Supporting ✔	This variant is absent from gnomAD v4.1.0 (PM2_Supporting)
PS4_Supporting ✔	This variant has been reported in one proband meeting an antithrombin activity level of < 0.8 IU/mL without confirmation of repeated independent samples tested, but has a family history of antithrombin activity levels of < 0.8 IU/mL (PS4_Supporting; PMID:11307839).

Curation History

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