Variant: NM_001386306.1:c.1097G>T

Version: 1.0

CA343772379

Gene: SERPINC1 (HGNC:462)
Condition: antithrombin III deficiency (MONDO:0013144)
Inheritance Mode: Autosomal dominant inheritance
UUID: f175fc11-74da-452a-a13b-adebd58169f1
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HGVS expressions

NM_001386306.1:c.1097G>T
NC_000001.11:g.173903971C>A
CM000663.2:g.173903971C>A
NC_000001.10:g.173873109C>A
CM000663.1:g.173873109C>A
NC_000001.9:g.172139732C>A
NG_012462.1:g.18408G>T
ENST00000367698.4:c.1313G>T
ENST00000367698.3:c.1313G>T
ENST00000617423.4:c.698G>T
NM_000488.3:c.1313G>T
NM_001365052.1:c.1169G>T
NM_000488.4:c.1313G>T
NM_001365052.2:c.1169G>T
NM_001386302.1:c.1436G>T
NM_001386303.1:c.1394G>T
NM_001386304.1:c.1292G>T
NM_001386305.1:c.1256G>T

Uncertain Significance

Met criteria codes 2
PP3 PM2_Supporting

Not Met criteria codes 2
PP4 BP4

Evidence Links 0

Expert Panel
Thrombosis VCEP

Criteria Specification Information
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Thrombosis VCEP

The c.1313G>T (NM_000488.3) variant in SERPINC1 is a missense variant predicted to cause substitution of arginine by methionine at amino acid 438 (p.Arg438Met). The variant is absent from gnomAD v2.1.1, v3.1, v4.0.0 with good coverage across both genomes and exomes, meeting criteria for PM2_supporting. The computational predictor REVEL gives a score of 0.82, which is above the threshold of

>0.6 and provides evidence that correlates with impact to SERPINC1 function, meeting criteria for PP3. In summary, based on the evidence available at this time, the clinical significance of this variant is uncertain. ACMG/AMP criteria applied, as specified by the Thrombosis Variant Curation Expert Panel for AT Deficiency for SERPINC1: PP3, PM2_Supporting.

Met criteria codes

PP3	✔	The computational predictor REVEL gives a score of 0.82, which is above the threshold of >0.6 and provides evidence that correlates with impact to SERPINC1 function (PP3).
PM2_Supporting	✔	The variant is absent from gnomAD v2.1.1, v3.1, and v4.0.0 with good coverage across both genomes and exomes, meeting criteria for PM2_supporting.

Not Met criteria codes

PP4	✖	No cases identified
BP4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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