

Variant: *NM_001114753.3(ENG):c.1762G>A (p.Val588Ile)*

Version: 1.0

CA5252632 [↗](#)

237024 (ClinVar) [↗](#)

Gene: ENG ([HGNC:2022](#))

Condition: telangiectasia, hereditary hemorrhagic, type 1
([MONDO:0008535](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: a0ff594f-5659-4165-814c-872b4d962ef0

Approved on: 2024/9/11

Published on: 2024/9/19

HGVS expressions

NM_001114753.3(ENG):c.1762G>A (p.Val588Ile)

NM_001114753.3:c.1762G>A

NC_000009.12:g.127816033C>T

CM000671.2:g.127816033C>T

NC_000009.11:g.130578312C>T

CM000671.1:g.130578312C>T

NC_000009.10:g.129618133C>T

NG_009551.1:g.43736G>A

NG_023245.1:g.18159C>T

ENST00000480266.6:c.1216G>A

ENST00000373203.9:c.1762G>A

ENST00000344849.4:c.1762G>A

ENST00000373203.8:c.1762G>A

ENST00000480266.5:c.1216G>A

NM_000118.3:c.1762G>A

NM_001114753.2:c.1762G>A

NM_001278138.1:c.1216G>A

NM_001278138.2:c.1216G>A

Likely Benign

Met criteria codes **2**

BP5 **BP4**

Not Met criteria codes **3**

BS1 **BA1** **PM2**

Evidence Links **0**

Expert Panel

Hereditary Hemorrhagic Telangiectasia VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** *ClinGen Hereditary Hemorrhagic Telangiectasia Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for ENG Version 1.1.0*

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

Hereditary Hemorrhagic Telangiectasia VCEP

The NM_001114753.3: c.1762G>A variant in ENG is a missense variant predicted to cause substitution of valine by isoleucine at amino acid 588 (p.Val588Ile). The highest population minor allele frequency in gnomAD v2.1.1 is 0.0003436 (43/125140 alleles) in the non-Finnish European population. The computational predictor REVEL gives a score of 0.043, which is below the threshold of ≤ 0.1 , and the splice site predictor SpliceAI indicated that the variant has no impact on splicing, evidence that does not predict a damaging effect on ENG function (BP4). This variant has been observed in a patient with an alternate molecular basis for disease (pathogenic variant identified in ACVRL1) (BP5; internal lab contributors). In summary, this variant meets the criteria to be classified as likely benign for autosomal dominant hereditary hemorrhagic telangiectasia based on the ACMG/AMP criteria applied, as specified by the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel: BP4, BP5 (specifications version 1.1.0; 09/11/2024).

Met criteria codes

BP5			This variant has been observed in a patient with an alternate molecular basis for disease (pathogenic variant identified in ACVRL1) (BP5; internal lab contributors).
BP4			The computational predictor REVEL gives a score of 0.043, which is below the threshold of ≤ 0.1 , and the splice site predictor SpliceAI indicated that the variant has no impact on splicing, evidence that does not predict a damaging effect on ENG function (BP4).

Not Met criteria codes

BS1			The highest population minor allele frequency in gnomAD v2.1.1 is 0.0003436 (43/125140 alleles) in the non-Finnish European population.
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



 

 

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