

Variant: *NM_001114753.3(ENG):c.1447G>A (p.Val483Ile)*

Version: 1.0

CA5252756 [↗](#)

414304 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 134b16b9-826a-4ab1-bdc9-6684cd6b7bcf

Approved on: 2025/2/4

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HGVS expressions

NM_001114753.3:c.1447G>A

NM_001114753.3(ENG):c.1447G>A (p.Val483Ile)

NC_000009.12:g.127818359C>T

CM000671.2:g.127818359C>T

NC_000009.11:g.130580638C>T

CM000671.1:g.130580638C>T

NC_000009.10:g.129620459C>T

NG_009551.1:g.41410G>A

ENST00000480266.6:c.901G>A

ENST00000373203.9:c.1447G>A

ENST00000344849.4:c.1447G>A

ENST00000373203.8:c.1447G>A

ENST00000480266.5:c.901G>A

NM_000118.3:c.1447G>A

NM_001114753.2:c.1447G>A

NM_001278138.1:c.901G>A

NR_136302.1:n.1426C>T

NM_001278138.2:c.901G>A

Likely Benign

Met criteria codes **2**

BP4

BS1_Supporting

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.1447G>A variant in ENG is a missense variant predicted to cause substitution of valine by isoleucine at amino acid 483 (p.Val483Ile). The filtering allele frequency (the lower threshold of the 95% CI of 28/19926) of the c.1447G>A variant in ENG is 0.0009914 for East Asian chromosomes by gnomAD v2.1.1, which is higher than the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel threshold (>0.0008-0.002) for BS1_Supporting, and therefore meets this criterion (BS1_Supporting). The computational predictor REVEL gives a score of 0.094, which is below the threshold of ≤0.15, 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). 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: BS1_Supporting, BP4 (specifications version 1.1.0; 02/04/2025).

Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.094, which is below the threshold of ≤0.15, 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).
BS1_Supporting			The filtering allele frequency (the lower threshold of the 95% CI of 28/19926) of the c.1447G>A variant in ENG is 0.0009914 for East Asian chromosomes by gnomAD v2.1.1, which is higher than the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel threshold (>0.0008-0.002) for BS1_Supporting, and therefore meets this criterion (BS1_Supporting).

Curation History



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