

Variant: *NM_001114753.3(ENG):c.1883C>T (p.Ala628Val)*

Version: 1.0

CA5252588 [↗](#)

1781928 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 330a5d6a-cf78-4282-ad59-6f92c9ae5c89

Approved on: 2026/4/10

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HGVS expressions

NM_001114753.3(ENG):c.1883C>T (p.Ala628Val)

NM_001114753.3:c.1883C>T

NC_000009.12:g.127815776G>A

CM000671.2:g.127815776G>A

NC_000009.11:g.130578055G>A

CM000671.1:g.130578055G>A

NC_000009.10:g.129617876G>A

NG_009551.1:g.43993C>T

NG_023245.1:g.17902G>A

ENST00000480266.6:c.1337C>T

ENST00000373203.9:c.1883C>T

ENST00000344849.4:c.*141C>T

ENST00000373203.8:c.1883C>T

ENST00000480266.5:c.1337C>T

NM_000118.3:c.*141C>T

NM_001114753.2:c.1883C>T

NM_001278138.1:c.1337C>T

NM_001278138.2:c.1337C>T

Likely Benign

Met criteria codes **2**

BP5 BP4

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.1883C>T variant in ENG is a missense variant predicted to cause substitution of alanine by valine at amino acid 628 (p.Ala628Val). The highest population minor allele frequency in gnomAD v2.1.1 is 0.00007858 (2/25452 alleles) in the Admixed American population. The computational predictor REVEL gives a score of 0.145, 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). 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; 04/10/2026).

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 0.145, 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).

Curation History



 

 

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