

Variant: *NM_001114753.3(ENG):c.920A>T (p.Asn307Ile)*

Version: 1.0

CA374982291 [↗](#)

1766194 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: d5ab7d77-ebad-4da7-bda8-55067d3641fa

Approved on: 2026/2/13

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HGVS expressions

NM_001114753.3(ENG):c.920A>T (p.Asn307Ile)

NM_001114753.3:c.920A>T

NC_000009.12:g.127824871T>A

CM000671.2:g.127824871T>A

NC_000009.11:g.130587150T>A

CM000671.1:g.130587150T>A

NC_000009.10:g.129626971T>A

NG_009551.1:g.34898A>T

ENST00000480266.6:c.374A>T

ENST00000373203.9:c.920A>T

ENST00000344849.4:c.920A>T

ENST00000373203.8:c.920A>T

ENST00000480266.5:c.374A>T

NM_000118.3:c.920A>T

NM_001114753.2:c.920A>T

NM_001278138.1:c.374A>T

NM_001278138.2:c.374A>T

Uncertain Significance

Met criteria codes **2**

PM2_Supporting

PS4_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.920A>T variant in ENG is a missense variant predicted to cause substitution of asparagine by isoleucine at amino acid 307 (p.Asn307Ile). This variant has not been observed in gnomAD v2.1 (PM2_Supporting) but has been observed in one individual with suspected HHT (PS4_Supporting). The computational predictor REVEL gives a score of 0.303, which is neither above nor below the thresholds predicting a damaging or benign impact on ENG function. In summary, this variant meets the criteria to be classified as a variant of uncertain significance 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: PM2_Supporting, PS4_Supporting (specifications version 1.1.0; 02/13/2026).

Met criteria codes

PM2_Supporting			This variant has not been observed in gnomAD v2.1 (PM2_Supporting).
PS4_Supporting			This variant has been observed in one individual with suspected HHT (PS4_Supporting).

Curation History



 

 

Showing 1 to 1 of 1 rows

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