

Variant: NM_001114753.3(ENG):c.1961C>G (p.Thr654Ser)

Version: 1.0

CA374969537 [↗](#)

426118 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: da64938b-7d69-47b6-b798-235f25be5a22

Approved on: 2024/3/15

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HGVS expressions

NM_001114753.3(ENG):c.1961C>G (p.Thr654Ser)

NM_001114753.3:c.1961C>G

NC_000009.12:g.127815698G>C

CM000671.2:g.127815698G>C

NC_000009.11:g.130577977G>C

CM000671.1:g.130577977G>C

NC_000009.10:g.129617798G>C

NG_009551.1:g.44071C>G

NG_023245.1:g.17824G>C

ENST00000480266.6:c.1415C>G

ENST00000373203.9:c.1961C>G

ENST00000344849.4:c.*219C>G

ENST00000373203.8:c.1961C>G

ENST00000480266.5:c.1415C>G

NM_000118.3:c.*219C>G

NM_001114753.2:c.1961C>G

NM_001278138.1:c.1415C>G

NM_001278138.2:c.1415C>G

Uncertain Significance

Met criteria codes **2**

BP4

PM2_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 1.0.0

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

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

Evidence submitted by expert panel

Hereditary Hemorrhagic Telangiectasia VCEP

The NM_001114753.3: c.1961C>G variant in ENG is a missense variant predicted to cause substitution of threonine by serine at amino acid 654 (p.Thr654Ser). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). The computational predictor REVEL gives a score of 0.126, 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). Due to limited evidence, this variant is 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, BP4 (specification version 1.0.0; 1/4/2024).

Met criteria codes

BP4			The computational predictor REVEL gives a score of 0.126, 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).
PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History



 

 

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