

Variant: *NM_001114753.3(ENG):c.1900T>G (p.Ser634Ala)*

Version: 1.0

CA200301183 [↗](#)

1782310 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: e762ece6-50cd-4694-b3b6-8ec9b6dca3ff

Approved on: 2026/4/10

Published on: 2026/8/28

HGVS expressions

NM_001114753.3:c.1900T>G

NM_001114753.3(ENG):c.1900T>G (p.Ser634Ala)

NC_000009.12:g.127815759A>C

CM000671.2:g.127815759A>C

NC_000009.11:g.130578038A>C

CM000671.1:g.130578038A>C

NC_000009.10:g.129617859A>C

NG_009551.1:g.44010T>G

NG_023245.1:g.17885A>C

ENST00000480266.6:c.1354T>G

ENST00000373203.9:c.1900T>G

ENST00000344849.4:c.*158T>G

ENST00000373203.8:c.1900T>G

ENST00000480266.5:c.1354T>G

NM_000118.3:c.*158T>G

NM_001114753.2:c.1900T>G

NM_001278138.1:c.1354T>G

NM_001278138.2:c.1354T>G

Uncertain Significance

Met criteria codes **1**

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 1.1.0

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

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

Evidence submitted by expert panel

Hereditary Hemorrhagic Telangiectasia VCEP

The NM_001114753.3: c.1900T>G variant in ENG is a missense variant predicted to cause substitution of serine by alanine at amino acid 634 (p.Ser634Ala). This variant has not been observed in gnomAD v2.1 (PM2_Supporting). The computational predictor REVEL gives a score of 0.158, 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 (specifications version 1.1.0; 04/10/2026).

Met criteria codes

PM2_Supporting ⓘ ✓ This variant has not been observed in gnomAD v2.1 (PM2_Supporting).

Curation History ⓘ



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