

Variant: *NM_001114753.3(ENG):c.392C>T (p.Pro131Leu)*

Version: 1.0

CA202695 [↗](#)

161232 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 007c246a-5626-4580-b6dc-c5817c428f04

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HGVS expressions

NM_001114753.3:c.392C>T

NM_001114753.3(ENG):c.392C>T (p.Pro131Leu)

NC_000009.12:g.127826641G>A

CM000671.2:g.127826641G>A

NC_000009.11:g.130588920G>A

CM000671.1:g.130588920G>A

NC_000009.10:g.129628741G>A

NG_009551.1:g.33128C>T

ENST00000480266.6:c.-155C>T

ENST00000373203.9:c.392C>T

ENST00000344849.4:c.392C>T

ENST00000373203.8:c.392C>T

ENST00000462196.1:n.292C>T

ENST00000480266.5:c.-155C>T

NM_000118.3:c.392C>T

NM_001114753.2:c.392C>T

NM_001278138.1:c.-155C>T

NM_001278138.2:c.-155C>T

Benign

Met criteria codes 1

BA1

Not Met criteria codes 1

PP3

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.392C>T variant in ENG is a missense variant predicted to cause substitution of proline by leucine at amino acid 131 (p.Pro131Leu). The filtering allele frequency (the lower threshold of the 95% CI of 762/30612) of the c.392C>T variant in ENG is 0.02343 for South Asian chromosomes by gnomAD v2.1.1, which is higher than the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel threshold (>0.01) for BA1, and therefore meets this criterion (BA1). The computational predictor REVEL gives a score of 0.315, 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 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: BA1 (specification version 1.0.0; 1/4/2024).

Met criteria codes

BA1			The filtering allele frequency (the lower threshold of the 95% CI of 762/30612) of the c.392C>T variant in ENG is 0.02343 for South Asian chromosomes by gnomAD v2.1.1, which is higher than the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel threshold (>0.01) for BA1, and therefore meets this criterion (BA1).
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Not Met criteria codes

PP3			The computational predictor REVEL gives a score of 0.315, which is neither above nor below the thresholds predicting a damaging or benign impact on ENG function.
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Curation History



 

 

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