Variant: NM_001114753.3(ENG):c.1807G>A (p.Gly603Arg)

Version: 1.1

CA374971982

995600 (ClinVar)

Gene: ENG (HGNC:2022)
Condition: telangiectasia, hereditary hemorrhagic, type 1 (MONDO:0008535)
Inheritance Mode: Autosomal dominant inheritance
UUID: 6086f2c6-c9b6-4fd8-b1fd-3342e29a7476
Approved on: 2024-11-12
Published on: 2024-11-15

HGVS expressions

NM_001114753.3:c.1807G>A
NM_001114753.3(ENG):c.1807G>A (p.Gly603Arg)
NC_000009.12:g.127815988C>T
CM000671.2:g.127815988C>T
NC_000009.11:g.130578267C>T
CM000671.1:g.130578267C>T
NC_000009.10:g.129618088C>T
NG_009551.1:g.43781G>A
NG_023245.1:g.18114C>T
ENST00000480266.6:c.1261G>A
ENST00000373203.9:c.1807G>A
ENST00000344849.4:c.1807G>A
ENST00000373203.8:c.1807G>A
ENST00000480266.5:c.1261G>A
NM_000118.3:c.1807G>A
NM_001114753.2:c.1807G>A
NM_001278138.1:c.1261G>A
NM_001278138.2:c.1261G>A

Likely Pathogenic

Met criteria codes 3

PS4 PP3 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.1807G>A variant in ENG is a missense variant predicted to cause substitution of glycine by arginine at amino acid 603 (p.Gly603Arg). This variant has been reported in 4 probands with a phenotype consistent with Hereditary Hemorrhagic Telangiectasia (PS4, Internal lab contributors, PMID: 20414677). This variant is absent from gnomAD v.2.1.1 (PM2_Supporting). The computational predictor REVEL gives a score of 0.876 which is above the threshold of 0.644, evidence that correlates with impact to ENG function (PP3). In summary, this variant meets the criteria to be classified as likely pathogenic 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: PS4, PM2_Supporting, PP3 (specifications version 1.1.0; 11/12/2024).

Met criteria codes

PS4	✔	This variant has been reported in 4 probands with a phenotype consistent with Hereditary Hemorrhagic Telangiectasia (PS4, Internal lab contributors, PMID: 20414677).
PP3	✔	The computational predictor REVEL gives a score of 0.876 which is above the threshold of 0.644, evidence that correlates with impact to ENG function (PP3).
PM2_Supporting	✔	This variant is absent from gnomAD v.2.1.1 (PM2_Supporting).

Curation History

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