

Variant: *NM_001114753.3(ENG):c.1701del (p.Val568fs)*

Version: 1.0

CA913190140 [↗](#)

618625 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 1faf3eb2-c488-4f8a-873e-9357828dcf78

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HGVS expressions

NM_001114753.3:c.1701del

NM_001114753.3(ENG):c.1701del (p.Val568fs)

NC_000009.12:g.127817189del

CM000671.2:g.127817189del

NC_000009.11:g.130579468del

CM000671.1:g.130579468del

NC_000009.10:g.129619289del

NG_009551.1:g.42580del

ENST00000480266.6:c.1155del

ENST00000373203.9:c.1701del

ENST00000344849.4:c.1701del

ENST00000373203.8:c.1701del

ENST00000480266.5:c.1155del

NM_000118.3:c.1701del

NM_001114753.2:c.1701del

NM_001278138.1:c.1155del

NR_136302.1:n.1124del

NM_001278138.2:c.1155del

Pathogenic

Met criteria codes **3**

PM2_Supporting

PVS1

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 1.0.0*

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

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

Evidence submitted by expert panel

Hereditary Hemorrhagic Telangiectasia VCEP

The NM_001114753.3: c.1701del (p.Val568Serfs*5) variant in ENG is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 13 (out of 15) leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). This variant has been reported in a proband with a phenotype consistent of HHT (PS4_Supporting; Internal lab contributors). In summary, this variant meets the criteria to be classified as 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: PVS1, PM2_Supporting, PS4_Supporting (specification version 1.0.0; 1/4/2024).

Met criteria codes

PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
PVS1			The NM_001114753.3: c.1701del (p.Val568Serfs*5) variant in ENG is a frameshift variant predicted to cause a premature stop codon in biologically relevant exon 13 (out of 15) leading to nonsense mediated decay in a gene in which loss-of-function is an established disease mechanism (PVS1).
PS4_Supporting			This variant has been reported in a proband with a phenotype consistent of HHT (PS4_Supporting; Internal lab contributors).

Curation History



 

 

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