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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
Approved on: 2024/3/15
Published on: 2024/3/15

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

PVS1 PM2_Supporting 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

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).
PM2_Supporting	✔	This variant is absent from gnomAD v2.1.1 (PM2_Supporting).
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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