

Variant: *NM_000020.3(ACVRL1):c.137G>C (p.Cys46Ser)*

Version: 1.0

CA384897664 [↗](#)

533345 (ClinVar) [↗](#)

Gene: ACVRL1 ([HGNC:94](#))

Condition: telangiectasia, hereditary hemorrhagic, type 2
([MONDO:0010880](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: e12475b5-168e-4263-8406-f945e2847af9

Approved on: 2024/1/4

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HGVS expressions

NM_000020.3(ACVRL1):c.137G>C (p.Cys46Ser)

NM_000020.3:c.137G>C

NC_000012.12:g.51913174G>C

CM000674.2:g.51913174G>C

NC_000012.11:g.52306958G>C

CM000674.1:g.52306958G>C

NC_000012.10:g.50593225G>C

NG_009549.1:g.10757G>C

ENST00000547400.6:c.179G>C

ENST00000551576.6:c.137G>C

ENST00000552678.2:c.137G>C

ENST00000388922.9:c.137G>C

ENST00000388922.8:c.137G>C

ENST00000419526.6:c.103+639G>C

ENST00000547400.5:c.179G>C

ENST00000550683.5:c.179G>C

ENST00000551576.5:c.137G>C

NM_000020.2:c.137G>C

NM_001077401.1:c.137G>C

NM_001077401.2:c.137G>C

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 ACVRL1 Version 1.1.0

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

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

Hereditary Hemorrhagic Telangiectasia VCEP

The NM_000020.3: c.137G>C variant in ACVRL1 is a missense variant predicted to cause substitution of cysteine by serine at amino acid 46 (p.Cys46Ser). This variant is absent from gnomAD v2.1.1 (PM2_Supporting). This variant has been reported in 4 probands with a phenotype consistent with HHT (PS4; ClinVar, Internal lab contributors). The computational predictor REVEL gives a score of 0.852, which is above the threshold of ≥ 0.644 , evidence that correlates with impact to ACVRL1 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 (specification version 1.1.0; 1/4/2024).

Met criteria codes

PS4			This variant has been reported in 4 probands with a phenotype consistent with HHT (PS4; ClinVar, Internal lab contributors).
PP3			The computational predictor REVEL gives a score of 0.852, which is above the threshold of ≥ 0.644 , evidence that correlates with impact to ACVRL1 function (PP3).
PM2_Supporting			This variant is absent from gnomAD v2.1.1 (PM2_Supporting).

Curation History

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