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Variant: NM_000020.3(ACVRL1):c.151T>G (p.Cys51Gly)

Version: 1.0

CA384897717

1381345 (ClinVar)

Gene: ACVRL1 (HGNC:94)
Condition: telangiectasia, hereditary hemorrhagic, type 2 (MONDO:0010880)
Inheritance Mode: Autosomal dominant inheritance
UUID: be352399-737d-4670-8f97-6e2043ab9fd9
Approved on: 2025/10/20
Published on: 2025/12/28

HGVS expressions

NM_000020.3:c.151T>G
NM_000020.3(ACVRL1):c.151T>G (p.Cys51Gly)
NC_000012.12:g.51913188T>G
CM000674.2:g.51913188T>G
NC_000012.11:g.52306972T>G
CM000674.1:g.52306972T>G
NC_000012.10:g.50593239T>G
NG_009549.1:g.10771T>G
ENST00000547400.6:c.193T>G
ENST00000551576.6:c.151T>G
ENST00000552678.2:c.151T>G
ENST00000388922.9:c.151T>G
ENST00000388922.8:c.151T>G
ENST00000419526.6:c.103+653T>G
ENST00000547400.5:c.193T>G
ENST00000550683.5:c.193T>G
ENST00000551576.5:c.151T>G
NM_000020.2:c.151T>G
NM_001077401.1:c.151T>G
NM_001077401.2:c.151T>G

Likely Pathogenic

Met criteria codes 4

PM5 PS4_Moderate PM2_Supporting 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 ACVRL1 Version 1.1.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

Hereditary Hemorrhagic Telangiectasia VCEP

The NM_000020.3: c.151T>G variant in ACVRL1 is a missense variant predicted to cause substitution of cysteine by glycine at amino acid 51 (p.Cys51Gly). The computational predictor REVEL gives a score of 0.917 which is above the threshold of 0.644, evidence that correlates with impact to ACVRL1 function (PP3). This variant is absent from gnomAD v.2.1.1 (PM2_Supporting). Another missense variant c.152G>A (p.Cys51Tyr) (PMIDs: 10694922, 16429404, 23722869) in the same codon has been classified as pathogenic for Hereditary Hemorrhagic Telangiectasia by the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel (PM5). This variant has been reported in 2 probands with a phenotype consistent with Hereditary Hemorrhagic Telangiectasia (PS4_Moderate, Ambry). 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: PM5, PS4_moderate, PM2_supporting, PP3 (specifications version 1.1.0; 10/20/2025).

Met criteria codes

PM5	✔	Another missense variant c.152G>A (p.Cys51Tyr) (PMIDs: 10694922, 16429404, 23722869) in the same codon has been classified as pathogenic for Hereditary Hemorrhagic Telangiectasia by the ClinGen Hereditary Hemorrhagic Telangiectasia Variant Curation Expert Panel (PM5).
PS4_Moderate	✔	This variant has been reported in 2 probands with a phenotype consistent with Hereditary Hemorrhagic Telangiectasia (PS4_Moderate, Ambry).
PM2_Supporting	✔	This variant is absent from gnomAD v.2.1.1 (PM2_Supporting).
PP3	✔	The computational predictor REVEL gives a score of 0.917 which is above the threshold of 0.644, evidence that correlates with impact to ACVRL1 function (PP3).

Curation History

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