The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000551.4(VHL):c.119C>T (p.Pro40Leu)

Version: 1.0

CA020046

135405 (ClinVar)

Gene: VHL (HGNC:7428)
Condition: von Hippel-Lindau disease (MONDO:0008667)
Inheritance Mode: Autosomal dominant inheritance
UUID: 3d606fc6-e2fd-4a23-8c42-ff8d668a7f9e
Approved on: 2024-06-25
Published on: 2024-07-17

HGVS expressions

NM_000551.4:c.119C>T
NM_000551.4(VHL):c.119C>T (p.Pro40Leu)
NC_000003.12:g.10141966C>T
CM000665.2:g.10141966C>T
NC_000003.11:g.10183650C>T
CM000665.1:g.10183650C>T
NC_000003.10:g.10158650C>T
NG_008212.3:g.5332C>T
ENST00000696142.1:c.119C>T
ENST00000696143.1:c.119C>T
ENST00000696153.1:c.119C>T
ENST00000256474.3:c.119C>T
ENST00000256474.2:c.119C>T
ENST00000345392.2:c.119C>T
NM_000551.3:c.119C>T
NM_198156.2:c.119C>T
NM_001354723.1:c.119C>T
NM_001354723.2:c.119C>T
NM_198156.3:c.119C>T

Benign

Met criteria codes 1

BA1

Evidence Links 0

Expert Panel

VHL VCEP

Criteria Specification Information

Criteria Specification: *ClinGen VHL Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for VHL Version 1.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

VHL VCEP

The variant NM_000551.4(VHL):c.119C>T (p.Pro40Leu) is a missense variant predicted to cause substitution of Proline by Leucine. The GroupMax Filtering Allele Frequency (95% CI) in gnomAD v4.1.0 is 0.0006831 (71/ 84504 from South Asian Population). This is higher than the ClinGen VHL VCEP threshold of >=0.000156 (0.0156%) threshold expected for VHL disease (BA1). In summary, this variant meets the criteria to be classified as Benign for autosomal-dominant von Hippel Lindau syndrome (VHL syndrome) based on the ACMG/AMP criteria applied, as specified by the ClinGen VHL VCEP Version 1.0 (Specifications approval date: 02/26/2024. Variant Approval Date 06/25/2024).

Met criteria codes

BA1	ℹ	✔	The GroupMax Filtering Allele Frequency (95% CI) in gnomAD v4.1.0 is 0.0006831 (71/ 84504 from South Asian Population). This is higher than the ClinGen VHL VCEP threshold of >=0.000156 (0.0156%) threshold expected for VHL disease (BA1).

Curation History

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