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.183C>G (p.Pro61=)

Version: 1.0

CA020075

135951 (ClinVar)

Gene: VHL (HGNC:7428)
Condition: von Hippel-Lindau disease (MONDO:0008667)
Inheritance Mode: Autosomal dominant inheritance
UUID: a119dbcc-dbab-408d-8997-31d27220a29f
Approved on: 2024-06-25
Published on: 2024-06-25

HGVS expressions

NM_000551.4:c.183C>G
NM_000551.4(VHL):c.183C>G (p.Pro61=)
NC_000003.12:g.10142030C>G
CM000665.2:g.10142030C>G
NC_000003.11:g.10183714C>G
CM000665.1:g.10183714C>G
NC_000003.10:g.10158714C>G
NG_008212.3:g.5396C>G
ENST00000696142.1:c.183C>G
ENST00000696143.1:c.183C>G
ENST00000696153.1:c.183C>G
ENST00000256474.3:c.183C>G
ENST00000256474.2:c.183C>G
ENST00000345392.2:c.183C>G
NM_000551.3:c.183C>G
NM_198156.2:c.183C>G
NM_001354723.1:c.183C>G
NM_001354723.2:c.183C>G
NM_198156.3:c.183C>G

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_000551.3(VHL):c.183C>G (p.Pro61=) is a silent variant in the first exon of VHL. The GroupMax Filtering Allele Frequency (95% CI) in gnomAD v4.1.0 is 0.001902 (192/89258 from South Asian Population). This is higher than the ClinGen VHL VCEP threshold of >=0.000156 (0.0156%) threshold expected for VHL disease (BA1) and is classified as Benign for autosomal-dominant von Hippel-Lindau disease (VHL disease) 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 0.001902 (192/89258 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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