

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: 2024/6/25

Published on: 2024/6/25

HGVS expressions

NM_000551.4(VHL):c.183C>G (p.Pro61=)

NM_000551.4:c.183C>G

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			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).
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Curation History



 

 

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