

Variant: *NM_000551.4(VHL):c.246C>T (p.Arg82=)*

Version: 1.0

CA020164 [↗](#)

137905 (ClinVar) [↗](#)

Gene: VHL ([HGNC:7428](#))

Condition: von Hippel-Lindau disease ([MONDO:0008667](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 4d337c56-d3e7-4abc-8cd9-ec05060ab40d

Approved on: 2024/6/25

Published on: 2024/6/25

HGVS expressions

NM_000551.4:c.246C>T

NM_000551.4(VHL):c.246C>T (p.Arg82=)

NC_000003.12:g.10142093C>T

CM000665.2:g.10142093C>T

NC_000003.11:g.10183777C>T

CM000665.1:g.10183777C>T

NC_000003.10:g.10158777C>T

NG_008212.3:g.5459C>T

ENST00000696142.1:c.246C>T

ENST00000696143.1:c.246C>T

ENST00000696153.1:c.246C>T

ENST00000256474.3:c.246C>T

ENST00000256474.2:c.246C>T

ENST00000345392.2:c.246C>T

NM_000551.3:c.246C>T

NM_198156.2:c.246C>T

NM_001354723.1:c.246C>T

NM_001354723.2:c.246C>T

NM_198156.3:c.246C>T

Benign

Met criteria codes **2**

BP4

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.246C>T (p.Arg82=) is a synonymous (silent) variant that does not display splicing impact according to SpliceAI and VarSeak (BP4). The GroupMax Filtering Allele Frequency (95% CI) in gnomAD v4.1.0 is 0.0002832 (365/1179692 from European, Non-Finnish 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

BP4			1) SpliceAI: 0 score 2) varSEAK: Class 1 (No splicing effect)
BA1			The GroupMax Filtering Allele Frequency (95% CI) in gnomAD v4.1.0 is 0.0002832 (365/1179692 from European, Non-Finnish 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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