

Variant: NM_000552.5(VWF):c.114C>T (p.Phe38=)

Version: 1.0

CA6403892 [↗](#)

310090 (ClinVar) [↗](#)

Gene: VWF ([HGNC:7450](#))

Condition: hereditary von Willebrand disease ([MONDO:0019565](#))

Inheritance Mode: Undetermined mode of inheritance

UUID: cfd48011-0567-4737-89f3-94111d00add9

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HGVS expressions

NM_000552.5:c.114C>T

NM_000552.5(VWF):c.114C>T (p.Phe38=)

NC_000012.12:g.6121280G>A

CM000674.2:g.6121280G>A

NC_000012.11:g.6230446G>A

CM000674.1:g.6230446G>A

NC_000012.10:g.6100707G>A

NG_009072.1:g.8391C>T

NG_009072.2:g.8391C>T

ENST00000261405.10:c.114C>T

ENST00000261405.9:c.114C>T

ENST00000321023.5:c.*173C>T

ENST00000538563.1:c.*173C>T

ENST00000538635.5:n.143C>T

NM_000552.3:c.114C>T

NM_000552.4:c.114C>T

Likely Benign

Met criteria codes **2**

BP7 BP4

Not Met criteria codes **2**

BP5 BS1

Evidence Links **0**

Expert Panel

[von Willebrand Disease VCEP](#) [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen von Willebrand Disease Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for VWF Version 1.0.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

von Willebrand Disease VCEP

The NM_000552.5:c.114C>T (p.Phe38=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP4 & BP7). A case carrying the c.114C>T (p.Phe38=) variant was also found to carry the ClinGen VWD VCEP curated pathogenic variant c.2435del. The

case is asserted to be VWD 3 which is typically a biallelic variant but there is not a second variant in this case curated pathogenic (PMID 16321553). Additionally, there are 8 submissions to ClinVar for this variant but for the submissions indicating affected status as yes, all classify the variant as likely benign. In summary, this variant meets the criteria to be classified as likely benign for hereditary von Willebrand disease based on the ACMG/AMP criteria applied, as specified by the ClinGen VWD VCEP: BP4, BP7 (VWD VCEP specifications v1l)

Met criteria codes

BP7			The c.114C>T (p.Phe38=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP7).
BP4			The c.114C>T (p.Phe38=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP4).

Not Met criteria codes

BP5			A case carrying the c.114C>T (p.Phe38=) variant was also found to carry the ClinGen VWD VCEP curated pathogenic variant c.2435del. The case is asserted to be VWD 3 which is typically a biallelic variant but there is not a second variant in this case curated pathogenic (PMID 16321553).
BS1			The Grpmax filtering allele frequency in gnomAD v4.1 is 0.004699 (based on 5669/1180026 alleles in the European non Finnish population, and 19 homozygotes). This intermediate allele frequency is lower than the ClinGen VWD VCEP threshold (>0.01) for BS1 but higher than the threshold (<0.0001 for type 2) for PM2_Supporting.

Curation History



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