

Variant: NM_000552.5(VWF):c.1077C>T (p.Pro359=)

Version: 1.0

CA6403568 [↗](#)

310086 (ClinVar) [↗](#)

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

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

Inheritance Mode: Undetermined mode of inheritance

UUID: ebf8f52f-ecdc-469b-9a2d-d41227f0945a

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HGVS expressions

NM_000552.5:c.1077C>T

NM_000552.5(VWF):c.1077C>T (p.Pro359=)

NC_000012.12:g.6072363G>A

CM000674.2:g.6072363G>A

NC_000012.11:g.6181529G>A

CM000674.1:g.6181529G>A

NC_000012.10:g.6051790G>A

NG_009072.1:g.57308C>T

NG_009072.2:g.57308C>T

ENST00000261405.10:c.1077C>T

ENST00000261405.9:c.1077C>T

ENST00000538635.5:n.420+38152C>T

NM_000552.3:c.1077C>T

NM_000552.4:c.1077C>T

Likely Benign

Met criteria codes **2**

BP7 BP4

Not Met criteria codes **1**

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.1077C>T (p.Pro359=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP4 & BP7). Three submitters have reported this variant to ClinVar but no phenotype details are available. There are no reports of affected cases in the literature and the variant is absent from LOVD. 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. (VCEP specifications version 1)

Met criteria codes

BP7			The NM_000552.5:c.1077C>T (p.Pro359=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP7).
BP4			The NM_000552.5:c.1077C>T (p.Pro359=) variant is a synonymous variant that is not predicted by SpliceAI to impact splicing (BP4).

Not Met criteria codes

BS1			The Grpmax filtering allele frequency in gnomAD v4.1 is 0.003637 (based on 362/91706 alleles in the south Asian population, this also includes 5 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.
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Curation History



 

 

Showing 1 to 1 of 1 rows

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