

Variant: NM_000314.8(PTEN):c.114T>G (p.Pro38=)

Version: 2.1

CA000116 [↗](#)

184068 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))

Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: a06a1116-f26c-4129-8a43-c1ecbda9bc38

Approved on: 2026/1/21

Published on: 2026/6/8

HGVS expressions

NM_000314.8:c.114T>G

NM_000314.8(PTEN):c.114T>G (p.Pro38=)

NC_000010.11:g.87894059T>G

CM000672.2:g.87894059T>G

NC_000010.10:g.89653816T>G

CM000672.1:g.89653816T>G

NC_000010.9:g.89643796T>G

NG_007466.2:g.35621T>G

ENST00000700029.2:c.114T>G

ENST00000710265.1:c.114T>G

ENST00000472832.3:c.114T>G

ENST00000688158.2:n.899+13621T>G

ENST00000688922.2:c.114T>G

ENST00000700021.1:c.114T>G

ENST00000700022.1:c.114T>G

ENST00000706954.1:c.114T>G

ENST00000706955.1:c.*149T>G

ENST00000686459.1:c.114T>G

ENST00000688158.1:c.*275+13621T>G

ENST00000688308.1:c.114T>G

ENST00000693560.1:c.633T>G

ENST00000371953.8:c.114T>G

ENST00000371953.7:c.114T>G

ENST00000462694.1:n.116T>G

ENST00000610634.1:c.12T>G

NM_000314.5:c.114T>G

NM_000314.6:c.114T>G

NM_001304717.2:c.633T>G

NM_001304718.1:c.-592T>G

NM_000314.7:c.114T>G

NM_001304717.5:c.633T>G

NM_001304718.2:c.-592T>G

Likely Benign

Met criteria codes 3

BP7BS3PM2_Supporting

Not Met criteria codes 23

PS1PS2PS3PS4BP5BP4BP3BP1BP2PP1PP2PP3PP4PM6BA1PM1PM3PM5PM4BS1BS4BS2PVS1

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specification: ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.2.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP			
PTEN c.114T>G (p.Pro38=) meets criteria to be classified as likely benign for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.1.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). BS3: Intronic OR synonymous variant with RNA, mini-gene, or other splicing assay demonstrating no splicing impact. (internal laboratory contributor Accession: SCV000213708.7) BP7: Variant is synonymous (silent) OR intronic and at or beyond +7/-21 and no splicing impact is predicted.			
Met criteria codes			
BP7			BP7: Variant is synonymous (silent) OR intronic and at or beyond +7/-21 and no splicing impact is predicted.
BS3			BS3: Intronic OR synonymous variant with RNA, mini-gene, or other splicing assay demonstrating no splicing impact. (internal laboratory contributor Accession: SCV000213708.7)
PM2_Supporting			The variant is present in gnomAD v4 with a frequency <0.00001 (0.00000183)
Not Met criteria codes			
PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			Splicing impact predicted by some tools - CONFLICTING. NNSplice no impact, MES no prediction, HSF potential cryptic site predicted.
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 		
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