

Variant: NM_000314.8(PTEN):c.1171C>T (p.Pro391Ser)

CA000118 [↗](#)

187673 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 2353f24f-8466-46ea-838d-a36a749f6a49

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HGVS expressions

NM_000314.8:c.1171C>T

NM_000314.8(PTEN):c.1171C>T (p.Pro391Ser)

NC_000010.11:g.87965431C>T

CM000672.2:g.87965431C>T

NC_000010.10:g.89725188C>T

CM000672.1:g.89725188C>T

NC_000010.9:g.89715168C>T

NG_007466.2:g.106993C>T

ENST00000700029.2:c.1264C>T

ENST00000710265.1:c.*200C>T

ENST00000688158.2:n.1906C>T

ENST00000688922.2:c.*1001C>T

ENST00000700021.1:c.1126C>T

ENST00000700022.1:c.*510C>T

ENST00000700023.1:n.2329C>T

ENST00000700024.1:n.2563C>T

ENST00000706954.1:c.1171C>T

ENST00000706955.1:c.*1206C>T

ENST00000686459.1:c.*757C>T

ENST00000688158.1:c.*1282C>T

ENST00000688308.1:c.1171C>T

ENST00000688922.1:c.1092C>T

ENST00000693560.1:c.1690C>T

ENST00000371953.8:c.1171C>T

ENST00000371953.7:c.1171C>T

NM_000314.5:c.1171C>T

NM_000314.6:c.1171C>T

NM_001304717.2:c.1690C>T

NM_001304718.1:c.580C>T

NM_000314.7:c.1171C>T

NM_001304717.5:c.1690C>T

NM_001304718.2:c.580C>T

Met criteria codes **4**

Uncertain Significance

BS3

BP5

PP2

PM2

Expert Panel

PTEN VCEP [↗](#)

Not Met criteria codes 22

- BA1
- BS4
- BS1
- BS2
- BP7
- BP3
- BP2
- BP1
- BP4
- PVS1
- PS4
- PS2
- PS1
- PS3
- PP4
- PP1
- PP3
- PM6
- PM5
- PM1
- PM4
- PM3

Evidence Links 0

Criteria Specification Information

[Criteria Specification:](#) *ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.1.0*

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

PTEN c.1171C>T (p.Pro391Ser) **PTEN c.1171C>T (p.Pro391Ser)** is currently classified as a variant of uncertain significance for **PTEN Hamartoma Tumor syndrome** in an autosomal dominant manner using modified ACMG criteria (PMID 30311380). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). **PM2: Present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533)** **PP2: PTEN is defined by the PTEN Expert Panel as a gene that has a low rate of benign missense variation and where missense variants are a common mechanism of disease. BS3: Missense variants with both lipid phosphatase activity AND results from a second assay appropriate to the protein domain demonstrating no statistically significant difference from wild type. (PMID 30993208, 29785012, 29706350)** **BS4_P: Lack of segregation in affected members of one family. (PMIDS: 28250423, 29706350, 29785012, 30993208, 32442409;internal laboratory contributor(s): SCV000218278.9,SCV000222170.9 and SCV000645550.8)**

Met criteria codes

BS3			Phosphatase activity, stability, pAKT, and subcellular localization similar to WT
BP5			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
PM2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Not Met criteria codes

BA1			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
BS1			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
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			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
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4			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
PS4			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
PS1			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
PP4			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
PP3			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
PM5			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
PM4			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
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Curation History ↗		
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