

Variant: *NM_000314.8(PTEN):c.287C>T (p.Pro96Leu)*

Version: 1.1

CA377482110 [↗](#)

449089 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 8f7f63bb-496b-4b77-8258-d706ccc91766

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HGVS expressions

NM_000314.8(PTEN):c.287C>T (p.Pro96Leu)

NM_000314.8:c.287C>T

NC_000010.11:g.87933046C>T

CM000672.2:g.87933046C>T

NC_000010.10:g.89692803C>T

CM000672.1:g.89692803C>T

NC_000010.9:g.89682783C>T

NG_007466.2:g.74608C>T

ENST00000700029.2:c.287C>T

ENST00000710265.1:c.287C>T

ENST00000472832.3:c.287C>T

ENST00000688158.2:n.1022C>T

ENST00000688922.2:c.*117C>T

ENST00000700021.1:c.242C>T

ENST00000700022.1:c.287C>T

ENST00000700029.1:c.121C>T

ENST00000706954.1:c.287C>T

ENST00000706955.1:c.*322C>T

ENST00000686459.1:c.287C>T

ENST00000688158.1:c.*398C>T

ENST00000688308.1:c.287C>T

ENST00000688922.1:c.208C>T

ENST00000693560.1:c.806C>T

ENST00000371953.8:c.287C>T

ENST00000371953.7:c.287C>T

ENST00000498703.1:n.113C>T

ENST00000610634.1:c.185C>T

NM_000314.5:c.287C>T

NM_000314.6:c.287C>T

NM_001304717.2:c.806C>T

NM_001304718.1:c.-464C>T

NM_000314.7:c.287C>T

NM_001304717.5:c.806C>T

NM_001304718.2:c.-464C>T

Likely Pathogenic

Met criteria codes 5

PS3_Moderate PP2 PP3 PM2_Supporting PM6

Not Met criteria codes 20

BA1 BP5 BP7 BP4 BP3 BP1 BP2 BS2 BS1 BS4 BS3 PP1 PP4 PM1 PM3 PM5 PM4 PS1 PS2 PS4

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 3.1.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.287C>T (p.Pro96Leu) meets criteria to be classified as likely pathogenic 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). PS3_Moderate: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -2.48 (≤ -1.11) on a high throughput phosphatase assay (PMID 29706350). PM2_Supporting: Absent in large sequenced populations (PMID 27535533). PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history (internal laboratory contributor SCV000616840.3). 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. PP3: REVEL score > 0.7 (score of this variant =0.933).

Met criteria codes

PS3_Moderate		✓	Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -2.13 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). Matreyek et al., variant is in possibly low abundance class - 0.48164 Mighell et al., score is -2.479382146 (TRUE) which is truncation like since less than -2.13
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.
PP3		✓	REVEL score > 0.7 (score of this variant = 0.933)
PM2_Supporting		✓	Absent in gnomAD
PM6		✓	Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (internal laboratory contributor: SCV000616840.3)

Not Met criteria codes

BA1		✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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BP5			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
BP4			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
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
BS2			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
BS3			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
PP4			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
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
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 

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