

Variant: *NM_000314.8(PTEN):c.113C>A (p.Pro38His)*

Version: 1.0

CA377784500 [↗](#)

1012092 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: fdcde17f-877a-4da6-8a12-e65ce252cbfa

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HGVS expressions

NM_000314.8(PTEN):c.113C>A (p.Pro38His)

NM_000314.8:c.113C>A

NC_000010.11:g.87894058C>A

CM000672.2:g.87894058C>A

NC_000010.10:g.89653815C>A

CM000672.1:g.89653815C>A

NC_000010.9:g.89643795C>A

NG_007466.2:g.35620C>A

ENST00000700029.2:c.113C>A

ENST00000710265.1:c.113C>A

ENST00000472832.3:c.113C>A

ENST00000688158.2:n.899+13620C>A

ENST00000688922.2:c.113C>A

ENST00000700021.1:c.113C>A

ENST00000700022.1:c.113C>A

ENST00000706954.1:c.113C>A

ENST00000706955.1:c.*148C>A

ENST00000686459.1:c.113C>A

ENST00000688158.1:c.*275+13620C>A

ENST00000688308.1:c.113C>A

ENST00000693560.1:c.632C>A

ENST00000371953.8:c.113C>A

ENST00000371953.7:c.113C>A

ENST00000462694.1:n.115C>A

ENST00000610634.1:c.11C>A

NM_000314.5:c.113C>A

NM_000314.6:c.113C>A

NM_001304717.2:c.632C>A

NM_001304718.1:c.-593C>A

NM_000314.7:c.113C>A

NM_001304717.5:c.632C>A

NM_001304718.2:c.-593C>A

Likely Pathogenic

Met criteria codes 4

PS2PM2_SupportingPS3_ModeratePM6

Not Met criteria codes 1

PM5

Evidence Links 1

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.113C>A (p.Pro38His) meets criteria to be classified as Pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.2.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor SCV005079984.1) PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (internal laboratory contributor SCV005079984.1) PS3_moderate: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-4.783403086] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PM2_supporting: Absent in large sequenced populations in the gnomAD v4 cohort. (PMID 27535533).

Met criteria codes

PS2		✓	De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor SCV005079984.1)
PM2_Supporting		✓	Absent in large sequenced populations in the gnomAD v4 cohort. (PMID 27535533).
PS3_Moderate		✓	Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-4.783403086] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350).
			Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-4.783403086] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PubMed:29706350
PM6		✓	Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (internal laboratory contributor SCV005079984.1)

Not Met criteria codes

PM5		✗	No other missense change determined to be pathogenic or likely pathogenic (by PTEN VCEP) and with equal or lesser BLOSUM62 score has been recorded in ClinVar.
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Curation History 

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