

Variant: *NM_000314.8(PTEN):c.278A>C (p.His93Pro)*

Version: 1.0

CA377482091 [↗](#)

468680 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 873374be-9923-42a8-9969-959167a621e8

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HGVS expressions

NM_000314.8:c.278A>C

NM_000314.8(PTEN):c.278A>C (p.His93Pro)

NC_000010.11:g.87933037A>C

CM000672.2:g.87933037A>C

NC_000010.10:g.89692794A>C

CM000672.1:g.89692794A>C

NC_000010.9:g.89682774A>C

NG_007466.2:g.74599A>C

ENST00000700029.2:c.278A>C

ENST00000710265.1:c.278A>C

ENST00000472832.3:c.278A>C

ENST00000688158.2:n.1013A>C

ENST00000688922.2:c.*108A>C

ENST00000700021.1:c.233A>C

ENST00000700022.1:c.278A>C

ENST00000700029.1:c.112A>C

ENST00000706954.1:c.278A>C

ENST00000706955.1:c.*313A>C

ENST00000686459.1:c.278A>C

ENST00000688158.1:c.*389A>C

ENST00000688308.1:c.278A>C

ENST00000688922.1:c.199A>C

ENST00000693560.1:c.797A>C

ENST00000371953.8:c.278A>C

ENST00000371953.7:c.278A>C

ENST00000498703.1:n.104A>C

ENST00000610634.1:c.176A>C

NM_000314.5:c.278A>C

NM_000314.6:c.278A>C

NM_001304717.2:c.797A>C

NM_001304718.1:c.-473A>C

NM_000314.7:c.278A>C

NM_001304717.5:c.797A>C

NM_001304718.2:c.-473A>C

Likely Pathogenic

Met criteria codes 6

- PM2_Supporting
- PP2
- PP3
- PM1
- PM5
- PS3_Moderate

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 3.0.0
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.278A>C (p.His93Pro) variant 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 3.0.0). 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 = -3.95 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PM1: Located at a residue within a catalytic motif as defined by the ClinGen PTEN Expert Panel. PM5: Missense change at an amino acid residue where a different missense change determined to be pathogenic or likely pathogenic and with equal or lesser BLOSUM62 score has been seen before (ClinVar Variation ID 7848, SCV000886857.1). PM2_Supporting: Absent in large sequenced populations (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. PP3: REVEL score > 0.7 (score of this variant =0.974).

Met criteria codes

PM2_Supporting			Absent in gnomAD
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=0.974)
PM1			Located at a residue within a catalytic motif as defined by the ClinGen PTEN Expert Panel (this variant locates at WPD Loop, residues 90-94)
PM5			Missense change at an amino acid residue where a different missense change determined to be pathogenic or likely pathogenic and with equal or lesser BLOSUM62 score has been seen before (ClinVar Variation ID: 7848). - BLOSUM62 scoreof this variant) = -2 - BLOSUM62 score of PTEN c.278A>G p.His93Arg = 0 [VCEP classification = PATH]
PS3_Moderate			Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -3.95 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350).
			Score = -3.95 (high_conf variant) PubMed:29706350 

Curation History



 

 

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