

Variant: *NM_000314.8(PTEN):c.213T>G (p.Cys71Trp)*

Version: 1.0

CA377481245 [↗](#)

820795 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 18913f2d-ee84-4f76-8417-78a493e61a72

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HGVS expressions

NM_000314.8:c.213T>G

NM_000314.8(PTEN):c.213T>G (p.Cys71Trp)

NC_000010.11:g.87931049T>G

CM000672.2:g.87931049T>G

NC_000010.10:g.89690806T>G

CM000672.1:g.89690806T>G

NC_000010.9:g.89680786T>G

NG_007466.2:g.72611T>G

ENST00000700029.2:c.213T>G

ENST00000710265.1:c.213T>G

ENST00000472832.3:c.213T>G

ENST00000688158.2:n.948T>G

ENST00000688922.2:c.*43T>G

ENST00000700021.1:c.168T>G

ENST00000700022.1:c.213T>G

ENST00000700029.1:c.47T>G

ENST00000706954.1:c.213T>G

ENST00000706955.1:c.*248T>G

ENST00000686459.1:c.213T>G

ENST00000688158.1:c.*324T>G

ENST00000688308.1:c.213T>G

ENST00000688922.1:c.134T>G

ENST00000693560.1:c.732T>G

ENST00000371953.8:c.213T>G

ENST00000371953.7:c.213T>G

ENST00000498703.1:n.39T>G

ENST00000610634.1:c.111T>G

NM_000314.5:c.213T>G

NM_000314.6:c.213T>G

NM_001304717.2:c.732T>G

NM_001304718.1:c.-538T>G

NM_000314.7:c.213T>G

NM_001304717.5:c.732T>G

NM_001304718.2:c.-538T>G

Likely Pathogenic

Met criteria codes 5

PM5

PP2

PP3

PM2_Supporting

PS3_Moderate

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.213T>G (p.Cys71Trp) 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 3.2.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column).

PM2_Supporting: Absent in large sequenced populations OR 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. PP3: REVEL score > 0.7 (score of this variant =0.946) PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-3.416566075] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). 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 936561).

Met criteria codes

PM5		✓	p.C71Y (c.212G>A) -> BLOSUM score -2 (Pathogenic variant by VCEP); p.C71W (c.213T>G) -> BLOSUM score -2 (this variant)
PP2		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3		✓	REVEL score 0.946 (>0.75)
PM2_Supporting		✓	Absent from gnomAD
PS3_Moderate		✓	Mighell et al score -3.416566075 (≤ -1.1)

Curation History

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