The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

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
Approved on: 2026/4/15
Published on: 2026/6/8

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

PM2_Supporting PP2 PP3 PM5 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

PM2_Supporting	✔	Absent from gnomAD
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)
PM5	✔	p.C71Y (c.212G>A) -> BLOSUM score -2 (Pathogenic variant by VCEP); p.C71W (c.213T>G) -> BLOSUM score -2 (this variant)
PS3_Moderate	✔	Mighell et al score –3.416566075 (<= -1.1)

Curation History

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