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.212G>A (p.Cys71Tyr)

Version: 1.0

CA377481238

936561 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 5236efa3-d087-480e-a42b-39335c9593d3
Approved on: 2024/2/9
Published on: 2024/3/4

HGVS expressions

NM_000314.8:c.212G>A
NM_000314.8(PTEN):c.212G>A (p.Cys71Tyr)
NC_000010.11:g.87931048G>A
CM000672.2:g.87931048G>A
NC_000010.10:g.89690805G>A
CM000672.1:g.89690805G>A
NC_000010.9:g.89680785G>A
NG_007466.2:g.72610G>A
ENST00000700029.2:c.212G>A
ENST00000710265.1:c.212G>A
ENST00000472832.3:c.212G>A
ENST00000688158.2:n.947G>A
ENST00000688922.2:c.*42G>A
ENST00000700021.1:c.167G>A
ENST00000700022.1:c.212G>A
ENST00000700029.1:c.46G>A
ENST00000706954.1:c.212G>A
ENST00000706955.1:c.*247G>A
ENST00000686459.1:c.212G>A
ENST00000688158.1:c.*323G>A
ENST00000688308.1:c.212G>A
ENST00000688922.1:c.133G>A
ENST00000693560.1:c.731G>A
ENST00000371953.8:c.212G>A
ENST00000371953.7:c.212G>A
ENST00000498703.1:n.38G>A
ENST00000610634.1:c.110G>A
NM_000314.5:c.212G>A
NM_000314.6:c.212G>A
NM_001304717.2:c.731G>A
NM_001304718.1:c.-539G>A
NM_000314.7:c.212G>A
NM_001304717.5:c.731G>A
NM_001304718.2:c.-539G>A

Pathogenic

Met criteria codes 5

PM2_Supporting | PP2 | PP3 | PS3_Moderate | PS2_Very Strong

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 3.0.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.212G>A (p.Cys71Tyr) 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 3.0.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PS2_VS: One proven plus two assumed de novo observations in a patient with the disease and no family history. (internal laboratory contributors: SCV003918479.1, SCV001376634.5) PS3_M: Functional studies showing a damaging effect on protein function. Phosphatase activity ≤ -1.11 per Mighell et al. 2018 (PMID: 29706350). This variant: score of -1.57581. 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 =0.957) PM2_P: Absent in the gnomAD cohort. (PMID 27535533).

Met criteria codes

PM2_Supporting	✔	Absent in gnomAD.
PP2	✔	Missense constraint
PP3	✔	Score of 0.957
PS3_Moderate	✔	Mighell et al. 2018 PMID: 29706350, Lipid phosphatase activity score, -1.57581 (TRUE). AND Han et al. 2000 PMID: 10866302, Deficient (similar to null) phosphatase activity (<50% of wild-type) reported in Fig 2 for C71Y.
PS2_Very Strong	✔	One proven plus two assumed de novo observations in a patient with the disease and no family history. (internal laboratory contributors: SCV003918479.1, SCV001376634.5)

Curation History

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