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.106G>C (p.Gly36Arg)

Version: 1.0

CA000283

189400 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 6978957a-8d30-4cb6-8f2a-f14e69a78254
Approved on: 2023/10/11
Published on: 2023/10/18

HGVS expressions

NM_000314.8:c.106G>C
NM_000314.8(PTEN):c.106G>C (p.Gly36Arg)
NC_000010.11:g.87894051G>C
CM000672.2:g.87894051G>C
NC_000010.10:g.89653808G>C
CM000672.1:g.89653808G>C
NC_000010.9:g.89643788G>C
NG_007466.2:g.35613G>C
ENST00000700029.2:c.106G>C
ENST00000710265.1:c.106G>C
ENST00000472832.3:c.106G>C
ENST00000688158.2:n.899+13613G>C
ENST00000688922.2:c.106G>C
ENST00000700021.1:c.106G>C
ENST00000700022.1:c.106G>C
ENST00000706954.1:c.106G>C
ENST00000706955.1:c.*141G>C
ENST00000686459.1:c.106G>C
ENST00000688158.1:c.*275+13613G>C
ENST00000688308.1:c.106G>C
ENST00000693560.1:c.625G>C
ENST00000371953.8:c.106G>C
ENST00000371953.7:c.106G>C
ENST00000462694.1:n.108G>C
ENST00000610634.1:c.4G>C
NM_000314.5:c.106G>C
NM_000314.6:c.106G>C
NM_001304717.2:c.625G>C
NM_001304718.1:c.-600G>C
NM_000314.7:c.106G>C
NM_001304717.5:c.625G>C
NM_001304718.2:c.-600G>C

Likely Pathogenic

Met criteria codes 5

PS3_Moderate | PS1_Moderate | PP2 | PP3 | PM2_Supporting

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.106G>C (p.Gly36Arg) 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). PS1_M: Same amino acid change as a previously established likely pathogenic variant regardless of nucleotide change (ClinVar Variation ID: 1334551) PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -2.69 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PM2_P: 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 = 0.995)

Met criteria codes

PS3_Moderate	✔	Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -2.69 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350).
PS1_Moderate	✔	Same amino acid change as a previously established likely pathogenic variant regardless of nucleotide change (ClinVar Variation ID: 1334551)
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 = 0.995) [In addition, SpliceAI shows weak SAS loss score 0.18 and some SAS gain score 0.27]
PM2_Supporting	✔	Absent in gnomAD

Curation History

Showing 1 to 1 of 1 rows

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