

Variant: NM_000314.8(PTEN):c.107G>A (p.Gly36Glu)

Version: 1.0

CA377784477 [↗](#)

486972 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 03ccb7a8-afaa-4bf5-8165-93fc607ae327

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HGVS expressions

NM_000314.8:c.107G>A

NM_000314.8(PTEN):c.107G>A (p.Gly36Glu)

NC_000010.11:g.87894052G>A

CM000672.2:g.87894052G>A

NC_000010.10:g.89653809G>A

CM000672.1:g.89653809G>A

NC_000010.9:g.89643789G>A

NG_007466.2:g.35614G>A

ENST00000700029.2:c.107G>A

ENST00000710265.1:c.107G>A

ENST00000472832.3:c.107G>A

ENST00000688158.2:n.899+13614G>A

ENST00000688922.2:c.107G>A

ENST00000700021.1:c.107G>A

ENST00000700022.1:c.107G>A

ENST00000706954.1:c.107G>A

ENST00000706955.1:c.*142G>A

ENST00000686459.1:c.107G>A

ENST00000688158.1:c.*275+13614G>A

ENST00000688308.1:c.107G>A

ENST00000693560.1:c.626G>A

ENST00000371953.8:c.107G>A

ENST00000371953.7:c.107G>A

ENST00000462694.1:n.109G>A

ENST00000610634.1:c.5G>A

NM_000314.5:c.107G>A

NM_000314.6:c.107G>A

NM_001304717.2:c.626G>A

NM_001304718.1:c.-599G>A

NM_000314.7:c.107G>A

NM_001304717.5:c.626G>A

NM_001304718.2:c.-599G>A

Pathogenic

Met criteria codes 6

PS3_Moderate

PS2

PP2

PP3

PM2_Supporting

PS4_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.107G>A (p.Gly36Glu) 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: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor: SCV001781083.1) PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.797438259 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor: SCV001781083.1) 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 = 0.98).

Met criteria codes

PS3_Moderate		✓	Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.797438259 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). Mighell et al. 2018 PMID: 29706350: Lipid phosphatase activity score, -1.797438259 (TRUE).
PS2		✓	De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor: SCV001781083.1)
PP2		✓	missense constraint
PP3		✓	REVEL score of 0.98 (>0.7)
PM2_Supporting		✓	Absent in gnomAD.
PS4_Supporting		✓	Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor: SCV001781083.1)

Curation History

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