

Variant: *NM_000314.8(PTEN):c.182A>G (p.His61Arg)*

Version: 1.0

CA000332 [↗](#)

189402 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 33bda7f3-e4dc-4319-b832-5fc864c6bd1e

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HGVS expressions

NM_000314.8(PTEN):c.182A>G (p.His61Arg)

NM_000314.8:c.182A>G

NC_000010.11:g.87925530A>G

CM000672.2:g.87925530A>G

NC_000010.10:g.89685287A>G

CM000672.1:g.89685287A>G

NC_000010.9:g.89675267A>G

NG_007466.2:g.67092A>G

ENST00000700029.2:c.182A>G

ENST00000710265.1:c.182A>G

ENST00000472832.3:c.182A>G

ENST00000688158.2:n.917A>G

ENST00000688922.2:c.182A>G

ENST00000700021.1:c.165-5516A>G

ENST00000700022.1:c.182A>G

ENST00000700029.1:c.16A>G

ENST00000706954.1:c.182A>G

ENST00000706955.1:c.*217A>G

ENST00000686459.1:c.182A>G

ENST00000688158.1:c.*293A>G

ENST00000688308.1:c.182A>G

ENST00000688922.1:c.51A>G

ENST00000693560.1:c.701A>G

ENST00000371953.8:c.182A>G

ENST00000371953.7:c.182A>G

ENST00000498703.1:n.8A>G

ENST00000610634.1:c.80A>G

NM_000314.5:c.182A>G

NM_000314.6:c.182A>G

NM_001304717.2:c.701A>G

NM_001304718.1:c.-541-5516A>G

NM_000314.7:c.182A>G

NM_001304717.5:c.701A>G

NM_001304718.2:c.-541-5516A>G

Likely Pathogenic

Met criteria codes 5

PS4_SupportingPP2PP3PM2_SupportingPS3_Moderate

Not Met criteria codes 3

PP1PM1PM5

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			
PTEN c.182A>G (p.His61Arg) meets criteria to be classified as likely pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (PMID 30311380). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PS3_M: Massively parallel functional assay interrogating phosphatase activity demonstrating a statistically significant difference from wild type (PMID: 29706350). PS4_P: Proband(s) with phenotype specificity score of 1-1.5 (PMID: 28526761). PM2_P: Absent in large sequenced populations (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 = 0.981.			
Met criteria codes			
PS4_Supporting			Hansen-Kiss (2017): 1 proband point for 17 yo M proband (see details below).
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 = 0.981)
PM2_Supporting			Absent in gnomAD
PS3_Moderate			Mighell 2018: high confidence variant, cum_score = -3.615
Not Met criteria codes			
PP1			1 Meiosis: Ambry #3: F in 50s w/ OFC 59.75 cm, thyroid adenoma @40s, breast ca @40s, 50s. Ambry #4 (son of Ambry #3): M in 30s w/ OFC 62 cm, ID.
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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