

Variant: NM_000314.6(PTEN):c.715A>G (p.Met239Val)

Version: 1.1

CA000189 [↗](#)

184878 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: dd41683a-d4b5-4e1a-aafb-d6cbe064561e

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HGVS expressions

NM_000314.6:c.715A>G

NM_000314.6(PTEN):c.715A>G (p.Met239Val)

NC_000010.11:g.87957933A>G

CM000672.2:g.87957933A>G

NC_000010.10:g.89717690A>G

CM000672.1:g.89717690A>G

NC_000010.9:g.89707670A>G

NG_007466.2:g.99495A>G

ENST00000700029.2:c.715A>G

ENST00000710265.1:c.715A>G

ENST00000472832.3:c.715A>G

ENST00000688158.2:n.1450A>G

ENST00000688922.2:c.*545A>G

ENST00000700021.1:c.670A>G

ENST00000700022.1:c.*54A>G

ENST00000700023.1:n.1873A>G

ENST00000700024.1:n.2107A>G

ENST00000700025.1:n.1484A>G

ENST00000700026.1:n.352A>G

ENST00000700029.1:c.549A>G

ENST00000706954.1:c.715A>G

ENST00000706955.1:c.*750A>G

ENST00000686459.1:c.*301A>G

ENST00000688158.1:c.*826A>G

ENST00000688308.1:c.715A>G

ENST00000688922.1:c.636A>G

ENST00000693560.1:c.1234A>G

ENST00000371953.8:c.715A>G

ENST00000371953.7:c.715A>G

ENST00000472832.2:c.142A>G

NM_000314.5:c.715A>G

NM_001304717.2:c.1234A>G

NM_001304718.1:c.124A>G

NM_000314.7:c.715A>G

NM_001304717.5:c.1234A>G

NM_001304718.2:c.124A>G

NM_000314.8:c.715A>G

Uncertain Significance

Met criteria codes 3

BP4 PP2 PM2_Supporting

Not Met criteria codes 23

PS1 PS2 PS3 PS4 BP5 BP7
BP3 BP1 BP2 PP1 PP3 PP4
PM6 BA1 PM1 PM3 PM5
PM4 BS1 BS4 BS3 BS2
PVS1

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.715A>G (p.Met239Val) is currently classified as a variant of uncertain significance 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). PM2_Supporting: Absent in large sequenced populations 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. BP4: REVEL score < 0.5 (score=0.421). Using the Bayesian point system (PMID: 29300386) for this variant with conflicting evidence: 1 benign supporting = -1. 2 pathogenic supporting = 2. Total = 1 (VUS).

Met criteria codes

BP4			REVEL score < 0.5 (score=0.421).
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.
PM2_Supporting			Absent in large sequenced populations in the gnomAD cohort. (PMID 27535533).

Not Met criteria codes

PS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			Not observed in population databases.
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3			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
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

BS1			Not observed in population databases.
BS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 	
	
Showing 1 to 2 of 2 rows	

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