

Variant: NM_000314.6(PTEN):c.389G>A (p.Arg130Gln)

CA000437 [↗](#)

7829 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 844aa93c-ba66-4b09-825a-8e5e0fb0a775

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HGVS expressions

NM_000314.6(PTEN):c.389G>A (p.Arg130Gln)

NM_000314.6:c.389G>A

NC_000010.11:g.87933148G>A

CM000672.2:g.87933148G>A

NC_000010.10:g.89692905G>A

CM000672.1:g.89692905G>A

NC_000010.9:g.89682885G>A

NG_007466.2:g.74710G>A

ENST00000700029.2:c.389G>A

ENST00000710265.1:c.389G>A

ENST00000472832.3:c.389G>A

ENST00000688158.2:n.1124G>A

ENST00000688922.2:c.*219G>A

ENST00000700021.1:c.344G>A

ENST00000700022.1:c.389G>A

ENST00000700029.1:c.223G>A

ENST00000706954.1:c.389G>A

ENST00000706955.1:c.*424G>A

ENST00000686459.1:c.389G>A

ENST00000688158.1:c.*500G>A

ENST00000688308.1:c.389G>A

ENST00000688922.1:c.310G>A

ENST00000693560.1:c.908G>A

ENST00000371953.8:c.389G>A

ENST00000371953.7:c.389G>A

ENST00000498703.1:n.215G>A

ENST00000610634.1:c.287G>A

NM_000314.5:c.389G>A

NM_001304717.2:c.908G>A

NM_001304718.1:c.-362G>A

NM_000314.7:c.389G>A

NM_001304717.5:c.908G>A

NM_001304718.2:c.-362G>A

NM_000314.8:c.389G>A

Pathogenic

Met criteria codes 6

PM6_Very Strong

PS4

PS3

PP2

PM1

PM2

Evidence Links 0

Expert Panel

Criteria Specification Information !

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

PTEN c.389G>A (p.R130Q) meets criteria to be classified as pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (Mester et al. 2018; manuscript in preparation). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PM6_VS: At least four assumed de novo observations in a patient with the disease and no family history. (PMID 22595938, PMID 22327138, internal laboratory contributor(s) SCV000222111.11) PS3: Phosphatase activity <50% of wild type (PMID 10866302) PS4: Probands with phenotype specificity score of 4-15.5 (PMID 26798346, PMID 17526801, PMID 23335809, PMID 22266152) PM1: Located at a residue within a catalytic motif as defined by the ClinGen PTEN Expert Panel. PM2: 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.

Met criteria codes

PM6_Very Strong		✓	Needs to be Very Strong level, but not currently an option in the VCI.
PS4		✓	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
PP2		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM1		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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