

Variant: *NM_000314.7(PTEN):c.389G>C (p.Arg130Pro)*

Version: 1.0

CA000439 [↗](#)

142018 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 10bca2c6-405c-4f3a-919f-53db4e026879

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HGVS expressions

NM_000314.7:c.389G>C

NM_000314.7(PTEN):c.389G>C (p.Arg130Pro)

NC_000010.11:g.87933148G>C

CM000672.2:g.87933148G>C

NC_000010.10:g.89692905G>C

CM000672.1:g.89692905G>C

NC_000010.9:g.89682885G>C

NG_007466.2:g.74710G>C

ENST00000700029.2:c.389G>C

ENST00000710265.1:c.389G>C

ENST00000472832.3:c.389G>C

ENST00000688158.2:n.1124G>C

ENST00000688922.2:c.*219G>C

ENST00000700021.1:c.344G>C

ENST00000700022.1:c.389G>C

ENST00000700029.1:c.223G>C

ENST00000706954.1:c.389G>C

ENST00000706955.1:c.*424G>C

ENST00000686459.1:c.389G>C

ENST00000688158.1:c.*500G>C

ENST00000688308.1:c.389G>C

ENST00000688922.1:c.310G>C

ENST00000693560.1:c.908G>C

ENST00000371953.8:c.389G>C

ENST00000371953.7:c.389G>C

ENST00000498703.1:n.215G>C

ENST00000610634.1:c.287G>C

NM_000314.5:c.389G>C

NM_000314.6:c.389G>C

NM_001304717.2:c.908G>C

NM_001304718.1:c.-362G>C

NM_001304717.5:c.908G>C

NM_001304718.2:c.-362G>C

NM_000314.8:c.389G>C

Pathogenic

Met criteria codes 5

PS4_Supporting

PS3

PP2

PM2

PM1

Not Met criteria codes 19

BA1

BS1

BS4

BS3

BS2

BP5

BP7

BP4

BP3

BP1

BP2

PS1

PS2

PP1

PP3

PP4

PM6

PM3

PM4

Evidence Links 1

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.389G>C (p.Arg130Pro) meets criteria to be classified as 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: Phosphatase activity <50% of wild type (PMID 29706350) 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. PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor SCV000185699.6)

Met criteria codes

PS4_Supporting

Proband in 60s with goiter, thyroid ca in 20s, kidney cancer in 60s, ganglioneuroma, >10 colon polyps, macrocephaly. (1 point) (Ambry Genetics).

PS3

High-throughput functional assay testing PTEN Lipid Phosphatase Activity. R130P had a score of -3.153431338 and was 'true' indicating function was truncation-like for PS3.

High-throughput functional assay testing PTEN Lipid Phosphatase Activity. R130P had a score of -3.153431338 and was 'true' indicating function was truncation-like for PS3.

PubMed:29706350

PP2

Missense.

PM2

Not present in gnomAD (coverage >90X).

PM1

Affects amino acid position 130, which is in a catalytic motif.

Not Met criteria codes

BA1

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

BS1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
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
BP4			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
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
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			Wrapped into PS4. Proband in 60s with goiter, thyroid ca in 20s, kidney cancer in 60s, ganglioneuroma, >10 colon polyps, macrocephaly. (1 point) (Ambry Genetics).

PM6			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
PM4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History 		
    		
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