

Variant: NM_000314.6(PTEN):c.44G>A (p.Arg15Lys)

CA16613142 [↗](#)

404147 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9b8faad8-e78a-4d2f-8805-210367466752

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HGVS expressions

NM_000314.6(PTEN):c.44G>A (p.Arg15Lys)

NM_000314.6:c.44G>A

NC_000010.11:g.87864513G>A

CM000672.2:g.87864513G>A

NC_000010.10:g.89624270G>A

CM000672.1:g.89624270G>A

NC_000010.9:g.89614250G>A

NG_007466.2:g.6075G>A

NG_033079.1:g.3925C>T

ENST00000700029.2:c.44G>A

ENST00000710265.1:c.44G>A

ENST00000472832.3:c.44G>A

ENST00000688922.2:c.44G>A

ENST00000700021.1:c.44G>A

ENST00000700022.1:c.44G>A

ENST00000706954.1:c.44G>A

ENST00000706955.1:c.44G>A

ENST00000686459.1:c.44G>A

ENST00000688158.1:c.44G>A

ENST00000688308.1:c.44G>A

ENST00000693560.1:c.563G>A

ENST00000371953.8:c.44G>A

ENST00000371953.7:c.44G>A

ENST00000462694.1:n.46G>A

ENST00000487939.1:n.65G>A

ENST00000610634.1:c.-59G>A

ENST00000618586.1:n.13G>A

NM_000314.5:c.44G>A

NM_001304717.2:c.563G>A

NM_001304718.1:c.-662G>A

NM_000314.7:c.44G>A

NM_001304717.5:c.563G>A

NM_001304718.2:c.-662G>A

NM_000314.8:c.44G>A

Likely Pathogenic

Met criteria codes 4

- PP2
- PM2_Supporting
- PM6_Strong
- PS3_Moderate

Not Met criteria codes 22

- PM5
- PM1
- PM4
- PM3
- BS2
- BS4
- BS3
- BS1
- PVS1
- PS1
- PS2
- PS4
- BP7
- BP5
- BP3
- BP2
- BP1
- BP4
- BA1
- PP4
- PP1
- PP3

Evidence Links 2

Expert Panel

PTEN VCEP

Criteria Specification Information

- [Criteria Specification:](#) ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.1.0
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.44G>A (p.Arg15Lys) meets criteria to be classified as likely pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.1.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PM2_P: Absent in large sequenced populations (PMID 27535533). PM6_S: One proband with presumed de novo occurrence (maternity/paternity not confirmed) for a patient with highly specific phenotype. (internal laboratory contributor: SCV000573362.4) 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. PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.65 ($\leq$ -1.11) on a high throughput phosphatase assay (PMID:29706350).

Met criteria codes

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).
PM6_Strong			One proband with presumed de novo occurrence (maternity/paternity not confirmed) for a patient with highly specific phenotype. (internal laboratory contributor: SCV000573362.4)
PS3_Moderate			Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.65 ($\leq$ -1.11) on a high throughput phosphatase assay (PMID:29706350).
Hypomorphic score -1.65 (meets based on updated classification rules if hypomorphic) PubMed:29706350 wt-like stability score 1.2 PubMed:29785012			

Not Met criteria codes

PM5			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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PM1			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
PM3			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
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
BS1			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
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
PS4			This data is used toward PM6_S.
BP7			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
BP3			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
BP1			

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
BA1			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
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			REVEL score of 0.7 (needs to be >0.7)

Curation History



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