The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

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
Approved on: 2018-01-03
Published on: 2018-12-10

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

PS3_Moderate PM6_Strong PM2_Supporting PP2

Not Met criteria codes 22

PM5 PM1 PM4 PM3 BA1 BS4 BS3 BS1 BS2 BP7 BP5 BP3 BP2 BP1 BP4 PVS1 PS1 PS2 PS4 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 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350).

Met criteria codes

PS3_Moderate	✔	Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.65 (≤ -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
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)
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.

Not Met criteria codes

PM5	✖	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
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
BA1		✖	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
BS2		✖	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
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
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.
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

Showing 1 to 2 of 2 rows

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