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.170T>G (p.Leu57Trp)

CA000131

185713 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: d0edcf69-04fa-4771-afb0-a49f8aab9ead
Approved on: 2018-04-06
Published on: 2018-12-10

HGVS expressions

NM_000314.6:c.170T>G
NM_000314.6(PTEN):c.170T>G (p.Leu57Trp)
NC_000010.11:g.87925518T>G
CM000672.2:g.87925518T>G
NC_000010.10:g.89685275T>G
CM000672.1:g.89685275T>G
NC_000010.9:g.89675255T>G
NG_007466.2:g.67080T>G
ENST00000700029.2:c.170T>G
ENST00000710265.1:c.170T>G
ENST00000472832.3:c.170T>G
ENST00000688158.2:n.905T>G
ENST00000688922.2:c.170T>G
ENST00000700021.1:c.165-5528T>G
ENST00000700022.1:c.170T>G
ENST00000700029.1:c.4T>G
ENST00000706954.1:c.170T>G
ENST00000706955.1:c.*205T>G
ENST00000686459.1:c.170T>G
ENST00000688158.1:c.*281T>G
ENST00000688308.1:c.170T>G
ENST00000688922.1:c.39T>G
ENST00000693560.1:c.689T>G
ENST00000371953.8:c.170T>G
ENST00000371953.7:c.170T>G
ENST00000610634.1:c.68T>G
NM_000314.5:c.170T>G
NM_001304717.2:c.689T>G
NM_001304718.1:c.-541-5528T>G
NM_000314.7:c.170T>G
NM_001304717.5:c.689T>G
NM_001304718.2:c.-541-5528T>G
NM_000314.8:c.170T>G

Met criteria codes 4

Expert Panel

Likely Pathogenic

PS3_Moderate PM2_Supporting PP2 PP3

Not Met criteria codes 22

PVS1 BS2 BS4 BS3 BS1 BP5 BP7 BP2 BP3 BP1 BP4 PM6 PM3 PM1 PM4 PM5 PS2 PS4 PS1 BA1 PP4 PP1

Evidence Links 2

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

PTEN c.170T>G (p.Leu57Trp) meets criteria to be classified as likely 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 9256433, 29706350) PM2: Absent in large sequenced populations 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

PS3_Moderate	✔	Matreyek et al. 2018 PMID: 29785012: abundance class was low PubMed:29785012 Score -4.86, high-confidence, truncation-like PubMed:29706350
PM2_Supporting	✔	Absent gnomAD
PP2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3	✔	REVEL score of 0.965 in UCSC Genome Browser since no score provided in the VCI

Not Met criteria codes

PVS1	✖	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	ℹ	✖	Absent gnomAD
BP5	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2	ℹ	✖	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
BP4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6	ℹ	✖	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
PM1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1	ℹ	✖	

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

BA1	✖	Absent gnomAD
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

Curation History

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