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.964A>T (p.Lys322Ter)

CA000235

185213 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: bfdfbf81-6f26-43f8-aa4b-bbb17ebaf54a
Approved on: 2017-10-18
Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.964A>T (p.Lys322Ter)
NM_000314.6:c.964A>T
NC_000010.11:g.87961056A>T
CM000672.2:g.87961056A>T
NC_000010.10:g.89720813A>T
CM000672.1:g.89720813A>T
NC_000010.9:g.89710793A>T
NG_007466.2:g.102618A>T
ENST00000700029.2:c.1057A>T
ENST00000710265.1:c.964A>T
ENST00000472832.3:c.964A>T
ENST00000688158.2:n.1699A>T
ENST00000688922.2:c.*794A>T
ENST00000700021.1:c.919A>T
ENST00000700022.1:c.*303A>T
ENST00000700023.1:n.2122A>T
ENST00000700024.1:n.2356A>T
ENST00000700025.1:n.1733A>T
ENST00000700026.1:n.601A>T
ENST00000706954.1:c.964A>T
ENST00000706955.1:c.*999A>T
ENST00000686459.1:c.*550A>T
ENST00000688158.1:c.*1075A>T
ENST00000688308.1:c.964A>T
ENST00000688922.1:c.885A>T
ENST00000693560.1:c.1483A>T
ENST00000371953.8:c.964A>T
ENST00000371953.7:c.964A>T
ENST00000472832.2:c.391A>T
NM_000314.5:c.964A>T
NM_001304717.2:c.1483A>T
NM_001304718.1:c.373A>T
NM_000314.7:c.964A>T
NM_001304717.5:c.1483A>T
NM_001304718.2:c.373A>T
NM_000314.8:c.964A>T

Likely Pathogenic

Met criteria codes 2

PM2 PVS1

Not Met criteria codes 23

PP3 PP2 PP1 PM5 PM1 PM4 PM3 BA1 PM6 BS2 BS4 BS3 BS1 PS4 PS2 PS1 PS3 BP3 BP2 BP4 BP1 BP7 BP5

Evidence Links 0

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

PTEN c.964A>T (p.Lys322Ter) 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). PVS1: Null variant predicted to result in nonsense-mediated decay or causing truncation/frameshift at or 5' to c.1121 (NM_000314.4). PM2: Absent in large sequenced populations

Met criteria codes

PM2	✔	PM2_P: Absent in gnomAD (v2 and v4)
PVS1	✔	PVS1: predicted to result in nonsense-mediated decay (NMD)

Not Met criteria codes

PP3	✖	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
PP1	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
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
PM6		✖	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
PS4		✖	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
PS1		✖	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
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
BP4		✖	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
BP7	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

Showing 1 to 3 of 3 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.