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_000314.8(PTEN):c.226_228del (p.Tyr76del)

Version: 1.0

CA470659984

436442 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 0bb863b4-5324-4fa2-ba3e-96c5e873bf25
Approved on: 2026/6/16
Published on: 2026/6/26

HGVS expressions

NM_000314.8(PTEN):c.226_228del (p.Tyr76del)
NM_000314.8:c.226_228del
NC_000010.11:g.87931062_87931064del
CM000672.2:g.87931062_87931064del
NC_000010.10:g.89690819_89690821del
CM000672.1:g.89690819_89690821del
NC_000010.9:g.89680799_89680801del
NG_007466.2:g.72624_72626del
ENST00000700029.2:c.226_228del
ENST00000710265.1:c.226_228del
ENST00000472832.3:c.226_228del
ENST00000688158.2:n.961_963del
ENST00000688922.2:c.*56_*58del
ENST00000700021.1:c.181_183del
ENST00000700022.1:c.226_228del
ENST00000700029.1:c.60_62del
ENST00000706954.1:c.226_228del
ENST00000706955.1:c.*261_*263del
ENST00000686459.1:c.226_228del
ENST00000688158.1:c.*337_*339del
ENST00000688308.1:c.226_228del
ENST00000688922.1:c.147_149del
ENST00000693560.1:c.745_747del
ENST00000371953.8:c.226_228del
ENST00000371953.7:c.226_228del
ENST00000498703.1:n.52_54del
ENST00000610634.1:c.124_126del
NM_000314.5:c.226_228del
NM_000314.6:c.226_228del
NM_001304717.2:c.745_747del
NM_001304718.1:c.-525_-523del
NM_000314.7:c.226_228del
NM_001304717.5:c.745_747del
NM_001304718.2:c.-525_-523del

Pathogenic

Met criteria codes 3

PM2_Supporting PS3_Moderate PS2_Very Strong

Not Met criteria codes 1

PM1

Evidence Links 1

Expert Panel

PTEN VCEP

Criteria Specification Information

- **Criteria Specification:** *ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.2.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

PTEN VCEP

PTEN c.226_228del (p.Tyr76del) meets criteria to be classified as Pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.2.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PS2_VS: At least two proven OR one proven plus two assumed de novo observations in a patient with the disease and no family history. (internal laboratory contributor GTR Lab ID: 26957). PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-1.456936006] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PM2_supporting: Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533).

Met criteria codes

PM2_Supporting	✔	Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533).
PS3_Moderate	✔	Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-1.456936006] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350).
		Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = [-1.456936006] (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PubMed:29706350
PS2_Very Strong	✔	At least two proven OR one proven plus two assumed de novo observations in a patient with the disease and no family history. (internal laboratory contributor GTR Lab ID: 26957).

Not Met criteria codes

PM1	✖	Not located at a residue within a catalytic motif as defined by the ClinGen PTEN Expert Panel.

Curation History

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See Report	Preferred Variant Title	Classification	Condition	Published Date	Version	Criteria Specification	Gene
View	NM_000314.8(PTEN):c.226_228del (p.T...	Pathogenic	PTEN Hamartoma Tumor Syndrome	2026-06-26	1.0	ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.2.0	PTEN

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