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.70G>C (p.Asp24His)

Version: 1.0

CA000187

186005 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 30c8d466-6d2e-4e86-857a-b4ae1d8244f7
Approved on: 2025/4/4
Published on: 2025/10/8

HGVS expressions

NM_000314.8:c.70G>C
NM_000314.8(PTEN):c.70G>C (p.Asp24His)
NC_000010.11:g.87864539G>C
CM000672.2:g.87864539G>C
NC_000010.10:g.89624296G>C
CM000672.1:g.89624296G>C
NC_000010.9:g.89614276G>C
NG_007466.2:g.6101G>C
NG_033079.1:g.3899C>G
ENST00000700029.2:c.70G>C
ENST00000710265.1:c.70G>C
ENST00000472832.3:c.70G>C
ENST00000688922.2:c.70G>C
ENST00000700021.1:c.70G>C
ENST00000700022.1:c.70G>C
ENST00000706954.1:c.70G>C
ENST00000706955.1:c.70G>C
ENST00000686459.1:c.70G>C
ENST00000688158.1:c.70G>C
ENST00000688308.1:c.70G>C
ENST00000693560.1:c.589G>C
ENST00000371953.8:c.70G>C
ENST00000371953.7:c.70G>C
ENST00000462694.1:n.72G>C
ENST00000487939.1:n.91G>C
ENST00000610634.1:c.-33G>C
ENST00000618586.1:n.39G>C
NM_000314.5:c.70G>C
NM_000314.6:c.70G>C
NM_001304717.2:c.589G>C
NM_001304718.1:c.-636G>C
NM_000314.7:c.70G>C
NM_001304717.5:c.589G>C
NM_001304718.2:c.-636G>C

Pathogenic

Met criteria codes 7

PM6 PS3_Moderate PS2 PP2 PP3 PM2_Supporting PS4_Supporting

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.70G>C (p.Asp24His) meets criteria to be classified as 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). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor(s) SCV005686053.1). PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (PMID 34374989). PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -3.602 (≤ -1.11) on a high throughput phosphatase assay (PMID 29706350). PP3: REVEL score > 0.7 (score of this variant = 0.98). 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. PS4_P: Proband(s) with phenotype specificity score of 1-1.5 (PMID 23764071). PM2_P: Absent in large sequenced populations (PMID 27535533).

Met criteria codes

PM6	✔	Saleh et al 2021: Assumed de novo in child (PED score = 4)
PS3_Moderate	✔	Functional evidence in two studies (Mighell et al 2018; Mingo et al 2018) in favor of pathogenic evidence.
PS2	✔	GeneDx: Confirmed de novo in child (Ped score = 4)
PP2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3	✔	REVEL score (0.98) > 0.7
PM2_Supporting	✔	gnomAD v2 (10-89624296-G-C): Absent gnomAD v4 (10-87864539-G-C): 1 total allele (AF in total pop 0.000068% < 0.001%)
PS4_Supporting	✔	Shuch et al 2013: Proband w/ CC score = 47 (1 proband pt).

Curation History

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