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.402G>C (p.Met134Ile)

Version: 1.1

CA377482340

619908 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 502914e3-3945-40a1-bd9c-032f106057a8
Approved on: 2025/8/1
Published on: 2025/10/8

HGVS expressions

NM_000314.8(PTEN):c.402G>C (p.Met134Ile)
NM_000314.8:c.402G>C
NC_000010.11:g.87933161G>C
CM000672.2:g.87933161G>C
NC_000010.10:g.89692918G>C
CM000672.1:g.89692918G>C
NC_000010.9:g.89682898G>C
NG_007466.2:g.74723G>C
ENST00000700029.2:c.402G>C
ENST00000710265.1:c.402G>C
ENST00000472832.3:c.402G>C
ENST00000688158.2:n.1137G>C
ENST00000688922.2:c.*232G>C
ENST00000700021.1:c.357G>C
ENST00000700022.1:c.402G>C
ENST00000700029.1:c.236G>C
ENST00000706954.1:c.402G>C
ENST00000706955.1:c.*437G>C
ENST00000686459.1:c.402G>C
ENST00000688158.1:c.*513G>C
ENST00000688308.1:c.402G>C
ENST00000688922.1:c.323G>C
ENST00000693560.1:c.921G>C
ENST00000371953.8:c.402G>C
ENST00000371953.7:c.402G>C
ENST00000498703.1:n.228G>C
ENST00000610634.1:c.300G>C
NM_000314.5:c.402G>C
NM_000314.6:c.402G>C
NM_001304717.2:c.921G>C
NM_001304718.1:c.-349G>C
NM_000314.7:c.402G>C
NM_001304717.5:c.921G>C
NM_001304718.2:c.-349G>C

Likely Pathogenic

Met criteria codes 5

PP1_Moderate | PS4_Supporting | PM2 | PP2 | PP3

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.402G>C (p.Met134Ile) 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). PM2: Absent in large sequenced populations (PMID 27535533). PP1_M: Co-segregation with disease in multiple affected family members, with 5 or 6 meioses observed. (PMID 23124040, internal laboratory contributor(s) ClinVar Organization ID: 26957) 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 23124040)

Met criteria codes

PP1_Moderate	✔	Also 1 meiosis from GeneDx family = 5 total
PS4_Supporting	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2	✔	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
PP3	✔	REVEL score of 0.892 per the UCSC Genome Browser

Curation History

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