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.113C>T (p.Pro38Leu)

Version: 1.0

CA377784501

818421 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 9083923e-5318-4eca-a618-b119cb117c46
Approved on: 2023/10/11
Published on: 2023/10/18

HGVS expressions

NM_000314.8:c.113C>T
NM_000314.8(PTEN):c.113C>T (p.Pro38Leu)
NC_000010.11:g.87894058C>T
CM000672.2:g.87894058C>T
NC_000010.10:g.89653815C>T
CM000672.1:g.89653815C>T
NC_000010.9:g.89643795C>T
NG_007466.2:g.35620C>T
ENST00000700029.2:c.113C>T
ENST00000710265.1:c.113C>T
ENST00000472832.3:c.113C>T
ENST00000688158.2:n.899+13620C>T
ENST00000688922.2:c.113C>T
ENST00000700021.1:c.113C>T
ENST00000700022.1:c.113C>T
ENST00000706954.1:c.113C>T
ENST00000706955.1:c.*148C>T
ENST00000686459.1:c.113C>T
ENST00000688158.1:c.*275+13620C>T
ENST00000688308.1:c.113C>T
ENST00000693560.1:c.632C>T
ENST00000371953.8:c.113C>T
ENST00000371953.7:c.113C>T
ENST00000462694.1:n.115C>T
ENST00000610634.1:c.11C>T
NM_000314.5:c.113C>T
NM_000314.6:c.113C>T
NM_001304717.2:c.632C>T
NM_001304718.1:c.-593C>T
NM_000314.7:c.113C>T
NM_001304717.5:c.632C>T
NM_001304718.2:c.-593C>T

Pathogenic

Met criteria codes 6

PS4_Supporting | PP2 | PP3 | PS2_Very Strong | PS3_Moderate | PM2_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 3.0.0*
- **Criteria Specification Approval History**
- **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.113C>T (p.Pro38Leu) 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 3.0.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PS2_VS: Two proven de novo observations in a patient with the disease and no family history. (internal laboratory contributor: SCV002757182.1). PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -3.772 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor: SCV002757182.1). PM2_P: Absent in gnomAD. 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. PP3: REVEL score > 0.7 (score of this variant = 0.923)

Met criteria codes

PS4_Supporting	✔	Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor: SCV002757182.1)
PP2	✔	missense constraint
PP3	✔	REVEL score of 0.923
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: SCV002757182.1)
PS3_Moderate	✔	Lipid phosphatase activity score of -3.772374452 (TRUE) in Mighell et al. 2018 PMID: 29706350.
PM2_Supporting	✔	Absent in gnomAD

Curation History

Showing 1 to 1 of 1 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.