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.209T>C (p.Leu70Pro)

CA000350

7826 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 12985d15-e1e7-45ab-b3e5-48e082086d4d
Approved on: 2018-11-28
Published on: 2018-12-10

HGVS expressions

NM_000314.8(PTEN):c.209T>C (p.Leu70Pro)
NM_000314.8:c.209T>C
NC_000010.11:g.87925557T>C
CM000672.2:g.87925557T>C
NC_000010.10:g.89685314T>C
CM000672.1:g.89685314T>C
NC_000010.9:g.89675294T>C
NG_007466.2:g.67119T>C
ENST00000700029.2:c.209T>C
ENST00000710265.1:c.209T>C
ENST00000472832.3:c.209T>C
ENST00000688158.2:n.944T>C
ENST00000688922.2:c.209T>C
ENST00000700021.1:c.165-5489T>C
ENST00000700022.1:c.209T>C
ENST00000700029.1:c.43T>C
ENST00000706954.1:c.209T>C
ENST00000706955.1:c.*244T>C
ENST00000686459.1:c.209T>C
ENST00000688158.1:c.*320T>C
ENST00000688308.1:c.209T>C
ENST00000688922.1:c.78T>C
ENST00000693560.1:c.728T>C
ENST00000371953.8:c.209T>C
ENST00000371953.7:c.209T>C
ENST00000498703.1:n.35T>C
ENST00000610634.1:c.107T>C
NM_000314.5:c.209T>C
NM_000314.6:c.209T>C
NM_001304717.2:c.728T>C
NM_001304718.1:c.-541-5489T>C
NM_000314.7:c.209T>C
NM_001304717.5:c.728T>C
NM_001304718.2:c.-541-5489T>C

Likely Pathogenic

Met criteria codes 5

PS3_Moderate PM2_Supporting PS4_Supporting PP2 PP3

Not Met criteria codes 21

PVS1 BS2 BS4 BS3 BS1 BP5 BP7 BP2 BP3 BP1 BP4 PM6 PM3 PM1 PM4 PM5 PS2 PS1 BA1 PP1 PP4

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.209T>C (p.Leu70Pro) meets criteria to be classified as likely pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.1.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PS3: Phosphatase activity <50% of wild type (PMID 29706350) PM2: Absent in large sequenced populations (PMID 27535533). 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. (internal laboratory contributor(s) ClinVar Organization ID: 26957)

Met criteria codes

PS3_Moderate	✔	Mighell et al data is approved for moderate weight in the truncation-like range
PM2_Supporting	✔	Absent gnomAD v2 and v4
PS4_Supporting	✔	Internal lab case ClinVar Organization ID: 26957 1 phenotype specificity point. Pt also published in 33887726
PP2	✔	Missense – PP2 met – Last NT but does not appear to impact splicing (low spliceAI scores)
PP3	✔	REVEL score 0.988

Not Met criteria codes

PVS1	✖	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		✖	Absent gnomAD
BP5		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7		✖	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
BP3		✖	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
BP4		✖	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
PM3		✖	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
PM5		✖	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

BA1	✖	Absent gnomAD
PP1	✖	1 meiosis internal lab case ClinVar Organization ID: 26957; proband infant F with macrocephaly, variant also present in brother with macro, lipoma, dev delay
PP4	✖	Moved to PS4. 3 internal lab cases: #1 Young adult F, eth unk, Macrocephaly, FTC dx late teens, goiter, lipoma dx 3. Min CC score = 21. #2: 40s Asian F, BrCA dx 40s, goiter dx late 20s. OFC not provided. CC score = 5. #3: Pediatric M, eth unk, Macrocephaly, lipoma, dev delay. Peds score = 5, 1 PP4 point. His baby sister with macrocephaly also positive: 1 meiosis

Curation History

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