

Variant: NM_000314.8(PTEN):c.651C>T (p.Val217=)

CA10587096 [↗](#)

255814 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))

Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 2f3d636c-4b73-48ee-93dd-243ce38e9778

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HGVS expressions

NM_000314.8(PTEN):c.651C>T (p.Val217=)

NM_000314.8:c.651C>T

NC_000010.11:g.87957869C>T

CM000672.2:g.87957869C>T

NC_000010.10:g.89717626C>T

CM000672.1:g.89717626C>T

NC_000010.9:g.89707606C>T

NG_007466.2:g.99431C>T

ENST00000700029.2:c.651C>T

ENST00000710265.1:c.651C>T

ENST00000472832.3:c.651C>T

ENST00000688158.2:n.1386C>T

ENST00000688922.2:c.*481C>T

ENST00000700021.1:c.606C>T

ENST00000700022.1:c.509C>T

ENST00000700023.1:n.1809C>T

ENST00000700024.1:n.2043C>T

ENST00000700025.1:n.1420C>T

ENST00000700026.1:n.288C>T

ENST00000700029.1:c.485C>T

ENST00000706954.1:c.651C>T

ENST00000706955.1:c.*686C>T

ENST00000686459.1:c.*237C>T

ENST00000688158.1:c.*762C>T

ENST00000688308.1:c.651C>T

ENST00000688922.1:c.572C>T

ENST00000693560.1:c.1170C>T

ENST00000371953.8:c.651C>T

ENST00000371953.7:c.651C>T

ENST00000472832.2:c.78C>T

NM_000314.5:c.651C>T

NM_000314.6:c.651C>T

NM_001304717.2:c.1170C>T

NM_001304718.1:c.60C>T

NM_000314.7:c.651C>T

NM_001304717.5:c.1170C>T

NM_001304718.2:c.60C>T

Likely Benign

Met criteria codes 3

BP7BP4PM2_Supporting

Not Met criteria codes 23

PM3PM1PM4PM5PM6BS2BS4BS3BS1PVS1PS2PS4PS3PS1BP5BP2BP3BP1BA1PP4PP1PP2PP3

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 3.2.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel			
PTEN VCEP			
PTEN c.651C>T (p.V217=) meets criteria to be classified as likely benign 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). PM2_Supporting: Absent in large sequenced populations (PMID 27535533). BP4: Synonymous variant where at least 2 out of 3 in silico models predict no splicing impact. BP7: Variant is synonymous (silent), nucleotide is not conserved, and no splicing impact is predicted.			
Met criteria codes			
BP7		✓	SpliceAI predicts no impact on splicing (Scores are <0.2) nor the creation of a new splice site. The nucleotide is not highly conserved (PhyloP 0.377).
BP4		✓	SpliceAI predicts no splicing impact (Scores <0.2 are considered evidence of benign.)
PM2_Supporting		✓	Absent in gnomAD v2 and v4.
Not Met criteria codes			
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

PM6			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 I agree (FH)
PVS1			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
PS4			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3			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
BP5			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
BA1			Absent gnomAD I agree (FH)
PP4			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

PP1			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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History



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