

Variant: NM_000314.7(PTEN):c.159A>G (p.Val53=)

Version: 2.0

CA000127 [↗](#)

184020 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))
Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))
Inheritance Mode: Autosomal dominant inheritance
UUID: 995c0e99-dc70-494f-bb25-ccabe14633fb
Approved on: 2024/2/9
Published on: 2024/3/4

HGVS expressions

NM_000314.7(PTEN):c.159A>G (p.Val53=)
NM_000314.7:c.159A>G
NC_000010.11:g.87894104A>G
CM000672.2:g.87894104A>G
NC_000010.10:g.89653861A>G
CM000672.1:g.89653861A>G
NC_000010.9:g.89643841A>G
NG_007466.2:g.35666A>G
ENST00000700029.2:c.159A>G
ENST00000710265.1:c.159A>G
ENST00000472832.3:c.159A>G
ENST00000688158.2:n.899+13666A>G
ENST00000688922.2:c.159A>G
ENST00000700021.1:c.159A>G
ENST00000700022.1:c.159A>G
ENST00000706954.1:c.159A>G
ENST00000706955.1:c.*194A>G
ENST00000686459.1:c.159A>G
ENST00000688158.1:c.*275+13666A>G
ENST00000688308.1:c.159A>G
ENST00000688922.1:c.28A>G
ENST00000693560.1:c.678A>G
ENST00000371953.8:c.159A>G
ENST00000371953.7:c.159A>G
ENST00000462694.1:n.161A>G
ENST00000610634.1:c.57A>G
NM_000314.5:c.159A>G
NM_000314.6:c.159A>G
NM_001304717.2:c.678A>G
NM_001304718.1:c.-547A>G
NM_001304717.5:c.678A>G
NM_001304718.2:c.-547A>G
NM_000314.8:c.159A>G

Likely Benign

BP4BS1_Supporting

PTEN VCEP

Not Met criteria codes 24

BS2PVS1BS4BS3BP3BP1BP2BP5BP7PS1PS2PS3PS4BA1PP1PP2PP3PP4PM1PM3PM5PM4PM6PM2

Evidence Links 0

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.159A>G (p.Val53=) 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 3.0.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). BS1_P: To be applied for variants with filtering allele frequency of 0.0000043 up to 0.000043 (0.00043% up to 0.0043%) in gnomAD. Popmax FAF of this variant=0.00004593 (9 alleles v4 gnomAD). BP4: Synonymous variant where SpliceAI model predict no splicing impact (acceptor gain 0.08, donor gain 0.13).

Met criteria codes

BP4			Synonymous variant where SpliceAI model predict no splicing impact (acceptor gain 0.08, donor gain 0.13).
BS1_Supporting			To be applied for variants with filtering allele frequency of 0.0000043 up to 0.000043 (0.00043% up to 0.0043%) in gnomAD. Popmax FAF of this variant=0.00004593 (9 alleles v4 gnomAD).

Not Met criteria codes

BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1			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
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
BP2		✖	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
BP7		✖	In silico models (MaxEnt and HSF) predict an alternate donor site will be strengthened.
PS1		✖	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
PS3		✖	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
BA1		✖	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		✖	In silico models (HSF, FF, MaxEnt) predict no deleterious impact on the native donor; however, MaxEnt and HSF predict an alternate donor site 5 NT upstream is strengthened.
PP4		✖	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
PM3		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

