

Variant: *NM_000314.8(PTEN):c.-862G>T*

Version: 1.2

CA000612 [↗](#)

7845 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: b7d1c7bd-f00c-4e8d-a6a5-e83fed03f5df

Approved on: 2025-06-06

Published on: 2025-10-08

HGVS expressions

NM_000314.8:c.-862G>T

NM_000314.8(PTEN):c.-862G>T

NC_000010.11:g.87863608G>T

CM000672.2:g.87863608G>T

NC_000010.10:g.89623365G>T

CM000672.1:g.89623365G>T

NC_000010.9:g.89613345G>T

NG_007466.2:g.5171G>T

NG_033079.1:g.4830C>A

ENST00000706954.1:c.-16-846G>T

ENST00000688308.1:c.-17+495G>T

ENST00000692337.1:c.50G>T

ENST00000693560.1:c.-342G>T

ENST00000371953.7:c.-862G>T

ENST00000610634.1:c.-964G>T

NM_000314.5:c.-861G>T

NM_000314.6:c.-861G>T

NM_001304717.2:c.-342G>T

NM_001304718.1:c.-1566G>T

Uncertain Significance

Met criteria codes **1**

PM2_Supporting

Not Met criteria codes **25**

PM1 PM3 PM5 BA1 PM4
PM6 BS1 BS4 BS3 BS2
BP5 BP7 BP4 BP3 BP1 BP2
PS1 PS2 PS3 PS4 PVS1
PP1 PP2 PP3 PP4

Evidence Links **1**

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**



PTEN VCEP


PTEN c.-861G>T (NC_000010.10:g.89623365G>T) is currently classified as a variant of uncertain significance 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).



Met criteria codes



PM2_Supporting   Absent in gnomAD (v2, v3, and v4)



Not Met criteria codes



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



PM5   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



PM4   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

BS1   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   Reduced promoter activity (to ~50% compared to wild type) in luciferase assay and reduced (20-40%) of wild type protein expression by Western blotting. [PubMed:17847000](#) 

BS2   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			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
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
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			Criteria not met based on review of functional data by expert panel. <hr/> Reduced promoter activity (to ~50% compared to wild type) in luciferase assay and reduced (20-40%) of wild type protein expression by Western blotting. PubMed:17847000 
PS4			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
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
PP4			

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

Curation History [↗](#)

Showing 1 to 3 of 3 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.