

Variant: *NM_000314.6(PTEN):c.766G>T (p.Glu256Ter)*

CA000569 [↗](#)

7828 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 4284ff17-816f-4321-a0b3-e3baeb0afc95

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HGVS expressions

NM_000314.6(PTEN):c.766G>T (p.Glu256Ter)

NM_000314.6:c.766G>T

NM_000314.5:c.766G>T

NM_001304717.2:c.1285G>T

NM_001304718.1:c.175G>T

NM_000314.7:c.766G>T

NM_001304717.5:c.1285G>T

NM_001304718.2:c.175G>T

ENST00000371953.7:c.766G>T

ENST00000472832.2:n.193G>T

NC_000010.11:g.87957984G>T

CM000672.2:g.87957984G>T

NC_000010.10:g.89717741G>T

CM000672.1:g.89717741G>T

NC_000010.9:g.89707721G>T

NG_007466.2:g.99546G>T

Pathogenic

Met criteria codes **4**

PM2

PM6

PS4_Supporting

PVS1

Not Met criteria codes **19**

BP7

BP5

BP4

BP2

PS1

PS3

PS2

PP3

PP2

PP1

PP4

PM5

PM4

PM1

BA1

BS1

BS3

BS4

BS2

Evidence Links **1**

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.766G>T (p.E256X) meets criteria to be classified as pathogenic 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). PVS1: Null variant predicted to result in nonsense-mediated decay or causing truncation/frameshift at or 5’ to c.1121 (NM_000314.4). PM2: Absent in large sequenced populations (PMID 27535533). PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (PMID 9832032) PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID 9832032)

Met criteria codes			
PM2		✓	Absent gnomAD
PM6		✓	PMID 9832032
PS4_Supporting		✓	PMID: 9832032
Variant in a 2yo male with macrocephaly (+4.5SD), 2 lipomas, psychomotor retardation. Peds score = 6, 1 PP4 point. PubMed:9832032 			
PVS1		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
Not Met criteria codes			
BP7		✗	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
BP4		✗	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
PS3		✗	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
PP3		✗	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
PP1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP4			Wrapped into PS4.
PM5			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
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1			Absent gnomAD
BS1			Absent gnomAD
BS3			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
BS2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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Showing 1 to 1 of 1 rows

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