

Variant: NM_000314.8(PTEN):c.1034T>C (p.Leu345Pro)

Version: 1.0

CA377487108 [↗](#)

484605 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 82136d77-950f-4f10-ad15-b80bd870a8ba

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HGVS expressions

NM_000314.8(PTEN):c.1034T>C (p.Leu345Pro)

NM_000314.8:c.1034T>C

NC_000010.11:g.87965294T>C

CM000672.2:g.87965294T>C

NC_000010.10:g.89725051T>C

CM000672.1:g.89725051T>C

NC_000010.9:g.89715031T>C

NG_007466.2:g.106856T>C

ENST00000700029.2:c.1127T>C

ENST00000710265.1:c.*63T>C

ENST00000688158.2:n.1769T>C

ENST00000688922.2:c.*864T>C

ENST00000700021.1:c.989T>C

ENST00000700022.1:c.*373T>C

ENST00000700023.1:n.2192T>C

ENST00000700024.1:n.2426T>C

ENST00000706954.1:c.1034T>C

ENST00000706955.1:c.*1069T>C

ENST00000686459.1:c.*620T>C

ENST00000688158.1:c.*1145T>C

ENST00000688308.1:c.1034T>C

ENST00000688922.1:c.955T>C

ENST00000693560.1:c.1553T>C

ENST00000371953.8:c.1034T>C

ENST00000371953.7:c.1034T>C

NM_000314.5:c.1034T>C

NM_000314.6:c.1034T>C

NM_001304717.2:c.1553T>C

NM_001304718.1:c.443T>C

NM_000314.7:c.1034T>C

NM_001304717.5:c.1553T>C

NM_001304718.2:c.443T>C

Pathogenic

Met criteria codes 5

PS4_Moderate

PS2

PS3_Supporting

PP2

PM2

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

[Criteria Specification:](#) ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

PTEN c.1034T>C (p.Leu345Pro) 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). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor(s) ClinVar Organization ID: 26957) PM2: Absent in large sequenced populations (PMID 27535533). PS4_M: Probands with phenotype specificity score of 2-3.5. (PMID 21659347, internal laboratory contributor(s) ClinVar Organization ID: 26957) 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. PS3_P: Abnormal in vitro cellular assay or transgenic model with phenotype different from wild type that does not meet PS3. (PMID 29785012, PMID 29706350)

Met criteria codes

PS4_Moderate	i	✓	1 point for internal GeneDx case, F child with macrocephaly, developmental delay, vascular malformation. Also 1 point for PMID 21659347 per VCEP discussion.
PS2	i	✓	GeneDx case
PS3_Supporting	i	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2	i	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2		✓	ABSENT

Curation History

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