

Variant: NM_000314.8(PTEN):c.116C>T (p.Ala39Val)

Version: 1.0

CA377784505 [↗](#)

2022313 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 5bdfc5aa-9b55-4409-97a6-03aeeaaf3601

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HGVS expressions

NM_000314.8(PTEN):c.116C>T (p.Ala39Val)

NM_000314.8:c.116C>T

NC_000010.11:g.87894061C>T

CM000672.2:g.87894061C>T

NC_000010.10:g.89653818C>T

CM000672.1:g.89653818C>T

NC_000010.9:g.89643798C>T

NG_007466.2:g.35623C>T

ENST00000700029.2:c.116C>T

ENST00000710265.1:c.116C>T

ENST00000472832.3:c.116C>T

ENST00000688158.2:n.899+13623C>T

ENST00000688922.2:c.116C>T

ENST00000700021.1:c.116C>T

ENST00000700022.1:c.116C>T

ENST00000706954.1:c.116C>T

ENST00000706955.1:c.*151C>T

ENST00000686459.1:c.116C>T

ENST00000688158.1:c.*275+13623C>T

ENST00000688308.1:c.116C>T

ENST00000693560.1:c.635C>T

ENST00000371953.8:c.116C>T

ENST00000371953.7:c.116C>T

ENST00000462694.1:n.118C>T

ENST00000610634.1:c.14C>T

NM_000314.5:c.116C>T

NM_000314.6:c.116C>T

NM_001304717.2:c.635C>T

NM_001304718.1:c.-590C>T

NM_000314.7:c.116C>T

NM_001304717.5:c.635C>T

NM_001304718.2:c.-590C>T

Likely Pathogenic

Met criteria codes 5

PS3_Moderate

PP2

PP3

PM2_Supporting

PS2

Not Met criteria codes 1

PS4

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 3.1.0*

[Criteria Specification Approval History](#)

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

PTEN c.116C>T (p.Ala39Val) meets criteria to be classified as Likely pathogenic 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). PS2: At least two proven OR one proven plus two assumed de novo observations in a patient with the disease and no family history. (SCV004169053.1). PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.74147277 ($\leq$ -1.11) on a high throughput phosphatase assay (PMID:29706350). 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. PP3: REVEL score > 0.7 (score of this variant =0.799). PM2_Supporting: Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533).

Met criteria codes

PS3_Moderate			Functional studies showing a damaging effect on protein function. Phosphatase activity $\leq$ -1.11 per Mighell et al. 2018 (PMID: 29706350). This variant: score of -1.74147277.
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.
PP3			In silico model predicts a splicing impact. (REVEL score = 0.799 > 0.75)
PM2_Supporting			Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533).
PS2			GeneDx #2 de novo via WES trio (ps2 fits) in female teen with macrocephaly and "distinctive facial features" NOS. Parents unaffected, no further details.

Not Met criteria codes

PS4			GeneDx #1 female teen with autism, ID, macrocephaly. Family members not tested, no further details. #1 Variant heterozygous in male child with macrocephaly, developmental delay, and autism.
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Curation History

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