

Variant: NM_000314.8(PTEN):c.30_32del (p.Ser10del)

Version: 1.0

CA645369491 [↗](#)

428224 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))
Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))
Inheritance Mode: Autosomal dominant inheritance
UUID: c5f9e697-a1e8-4332-aa58-6954a9570db6
Approved on: 2026/6/16
Published on: 2026/6/26

HGVS expressions

NM_000314.8:c.30_32del
NM_000314.8(PTEN):c.30_32del (p.Ser10del)
NC_000010.11:g.87864499_87864501del
CM000672.2:g.87864499_87864501del
NC_000010.10:g.89624256_89624258del
CM000672.1:g.89624256_89624258del
NC_000010.9:g.89614236_89614238del
NG_007466.2:g.6061_6063del
NG_033079.1:g.3939_3941del
ENST00000700029.2:c.30_32del
ENST00000710265.1:c.30_32del
ENST00000472832.3:c.30_32del
ENST00000688922.2:c.30_32del
ENST00000700021.1:c.30_32del
ENST00000700022.1:c.30_32del
ENST00000706954.1:c.30_32del
ENST00000706955.1:c.30_32del
ENST00000686459.1:c.30_32del
ENST00000688158.1:c.30_32del
ENST00000688308.1:c.30_32del
ENST00000693560.1:c.549_551del
ENST00000371953.8:c.30_32del
ENST00000371953.7:c.30_32del
ENST00000462694.1:n.32_34del
ENST00000487939.1:n.51_53del
ENST00000610634.1:c.-73_-71del
NM_000314.5:c.30_32del
NM_000314.6:c.30_32del
NM_001304717.2:c.549_551del
NM_001304718.1:c.-676_-674del
NM_000314.7:c.30_32del
NM_001304717.5:c.549_551del
NM_001304718.2:c.-676_-674del

Uncertain Significance

PS3_Moderate

PM6

PM2_Supporting

Not Met criteria codes 1

PM4

Evidence Links 0

PTEN VCEP

Criteria Specification Information

Criteria Specification:

 ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.2.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.30_32del (p.Ser10del) is currently classified as a variant of uncertain significance for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.2.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PM2_Supporting: Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort. (PMID 27535533). PS3_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -4.623 (≤ -1.11) on a high throughput phosphatase assay (PMID:29706350) PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. (internal laboratory contributor(s) SCV000580011.6)

Met criteria codes

PS3_Moderate

i

✓

Mighell et al score of -4.628232038

PM6

i

✓

Assumed de novo mutation in one individual with epilepsy, developmental delay with regression, and macrocephaly (SSCV000580011.6)

PM2_Supporting

i

✓

Absent from gnomAD v2 and v4

Not Met criteria codes

PM4

i

✗

Not in catalytic domain

Curation History

▾

▾

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