

Variant: NM_000314.8(PTEN):c.400A>G (p.Met134Val)

Version: 1.0

CA377482334 [↗](#)

2033861 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: c8832939-b814-49d6-8fd9-c35f3d40adbc

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HGVS expressions

NM_000314.8(PTEN):c.400A>G (p.Met134Val)

NM_000314.8:c.400A>G

NC_000010.11:g.87933159A>G

CM000672.2:g.87933159A>G

NC_000010.10:g.89692916A>G

CM000672.1:g.89692916A>G

NC_000010.9:g.89682896A>G

NG_007466.2:g.74721A>G

ENST00000700029.2:c.400A>G

ENST00000710265.1:c.400A>G

ENST00000472832.3:c.400A>G

ENST00000688158.2:n.1135A>G

ENST00000688922.2:c.*230A>G

ENST00000700021.1:c.355A>G

ENST00000700022.1:c.400A>G

ENST00000700029.1:c.234A>G

ENST00000706954.1:c.400A>G

ENST00000706955.1:c.*435A>G

ENST00000686459.1:c.400A>G

ENST00000688158.1:c.*511A>G

ENST00000688308.1:c.400A>G

ENST00000688922.1:c.321A>G

ENST00000693560.1:c.919A>G

ENST00000371953.8:c.400A>G

ENST00000371953.7:c.400A>G

ENST00000498703.1:n.226A>G

ENST00000610634.1:c.298A>G

NM_000314.5:c.400A>G

NM_000314.6:c.400A>G

NM_001304717.2:c.919A>G

NM_001304718.1:c.-351A>G

NM_000314.7:c.400A>G

NM_001304717.5:c.919A>G

NM_001304718.2:c.-351A>G

Pathogenic

Met criteria codes 6

PM2_SupportingPS3_ModeratePS2PP2PP3PM6

Not Met criteria codes 2

BP1PM1

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 3.2.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.400A>G (p.Met134Val) 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 3.2.0). 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. PM6: Assumed de novo, but without confirmation of paternity and maternity in a patient with the disease and no family history. PS4: Proband(s) with phenotype specificity score of 2-3.5. (PMID 40564777). 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 0.75(score of this variant =0.86). PM2_P: 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

PM2_Supporting

Absent from gnomAD

PS3_Moderate

Mighell et al = -1.608 < -1.1

PS2

Confirmed de novo patient with macrocephaly and motor delay

PP2

missense variant

PP3

REVEL score 0.86 >0.75

PM6

assumed de novo patient with Macrocephaly and developmental delay

Not Met criteria codes

BP1

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

Curation History [↗](#)

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