

Variant: NM_000314.8(PTEN):c.373A>G (p.Lys125Glu)

Version: 1.0

CA377482291 [↗](#)

1338262 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 070c03ed-e8a2-458a-b3aa-2b37d8e9b93b

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HGVS expressions

NM_000314.8(PTEN):c.373A>G (p.Lys125Glu)

NM_000314.8:c.373A>G

NC_000010.11:g.87933132A>G

CM000672.2:g.87933132A>G

NC_000010.10:g.89692889A>G

CM000672.1:g.89692889A>G

NC_000010.9:g.89682869A>G

NG_007466.2:g.74694A>G

ENST00000700029.2:c.373A>G

ENST00000710265.1:c.373A>G

ENST00000472832.3:c.373A>G

ENST00000688158.2:n.1108A>G

ENST00000688922.2:c.*203A>G

ENST00000700021.1:c.328A>G

ENST00000700022.1:c.373A>G

ENST00000700029.1:c.207A>G

ENST00000706954.1:c.373A>G

ENST00000706955.1:c.*408A>G

ENST00000686459.1:c.373A>G

ENST00000688158.1:c.*484A>G

ENST00000688308.1:c.373A>G

ENST00000688922.1:c.294A>G

ENST00000693560.1:c.892A>G

ENST00000371953.8:c.373A>G

ENST00000371953.7:c.373A>G

ENST00000498703.1:n.199A>G

ENST00000610634.1:c.271A>G

NM_000314.5:c.373A>G

NM_000314.6:c.373A>G

NM_001304717.2:c.892A>G

NM_001304718.1:c.-378A>G

NM_000314.7:c.373A>G

NM_001304717.5:c.892A>G

NM_001304718.2:c.-378A>G

Pathogenic

Met criteria codes 6

PS2

PP2

PP3

PS3_Moderate

PM1

PM2_Supporting

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.373A>G (p.Lys125Glu) 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) SCV003642767.2). PM1: Located at a residue within a catalytic motif as defined by the ClinGen PTEN Expert Panel. PS3_M: Multiple functional studies supportive of a damaging effect on the gene or gene product (PMIDs 29706350, 19457929, 21828076, 20926450, 23475934). PP3: REVEL score > 0.7 (score of this variant = 0.951). 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. PM2_P: Absent in large sequenced populations (PMID 27535533).

Met criteria codes

PS2			Confirmed de novo in an Ambry proband (Ped score = 4)
PP2			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3			REVEL score (0.951) > 0.7
PS3_Moderate			Multiple functional studies with concordant evidence in the pathogenic direction.
PM1			P-loop (phosphatase core, residues 123-130)
PM2_Supporting			Absent in gnomAD v2 and v4

Curation History

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