

Variant: NM_000314.8(PTEN):c.1021T>G (p.Phe341Val)

Version: 1.0

CA377486165 [↗](#)

495806 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 3cd03060-6fba-4693-a164-a18c3b6fb681

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HGVS expressions

NM_000314.8(PTEN):c.1021T>G (p.Phe341Val)

NM_000314.8:c.1021T>G

NC_000010.11:g.87961113T>G

CM000672.2:g.87961113T>G

NC_000010.10:g.89720870T>G

CM000672.1:g.89720870T>G

NC_000010.9:g.89710850T>G

NG_007466.2:g.102675T>G

ENST00000700029.2:c.1114T>G

ENST00000710265.1:c.1021T>G

ENST00000472832.3:c.1021T>G

ENST00000688158.2:n.1756T>G

ENST00000688922.2:c.*851T>G

ENST00000700021.1:c.976T>G

ENST00000700022.1:c.*360T>G

ENST00000700023.1:n.2179T>G

ENST00000700024.1:n.2413T>G

ENST00000700025.1:n.1790T>G

ENST00000700026.1:n.658T>G

ENST00000706954.1:c.1021T>G

ENST00000706955.1:c.*1056T>G

ENST00000686459.1:c.*607T>G

ENST00000688158.1:c.*1132T>G

ENST00000688308.1:c.1021T>G

ENST00000688922.1:c.942T>G

ENST00000693560.1:c.1540T>G

ENST00000371953.8:c.1021T>G

ENST00000371953.7:c.1021T>G

ENST00000472832.2:c.448T>G

NM_000314.5:c.1021T>G

NM_000314.6:c.1021T>G

NM_001304717.2:c.1540T>G

NM_001304718.1:c.430T>G

NM_000314.7:c.1021T>G

NM_001304717.5:c.1540T>G

NM_001304718.2:c.430T>G

Uncertain Significance

Met criteria codes 4

PP2PP3PM2PS3_Supporting

Not Met criteria codes 1

BS3

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.1021T>G (p.Phe341Val) 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 3.1.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). 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_P: REVEL score > 0.7 (score of this variant =0.909) PM2_Sup:Absent in large sequenced populations

Met criteria codes

PP2

i

✓

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

i

✓

REVEL score is .909

PM2

✓

Absent in population

PS3_Supporting

i

✓

Mighell et al- score is .18 PS3_Sup

Not Met criteria codes

BS3

i

✗

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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