The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000314.8(PTEN):c.521A>G (p.Tyr174Cys)

CA349032

220007 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 3905138e-990e-4b30-b0aa-96ad556248ac
Approved on: 2018-09-26
Published on: 2018-12-10

HGVS expressions

NM_000314.8(PTEN):c.521A>G (p.Tyr174Cys)
NM_000314.8:c.521A>G
NC_000010.11:g.87952146A>G
CM000672.2:g.87952146A>G
NC_000010.10:g.89711903A>G
CM000672.1:g.89711903A>G
NC_000010.9:g.89701883A>G
NG_007466.2:g.93708A>G
ENST00000686459.1:c.*107A>G
ENST00000688158.1:c.*632A>G
ENST00000688308.1:c.521A>G
ENST00000688922.1:c.442A>G
ENST00000693560.1:c.1040A>G
ENST00000371953.8:c.521A>G
ENST00000371953.7:c.521A>G
NM_000314.5:c.521A>G
NM_000314.6:c.521A>G
NM_001304717.2:c.1040A>G
NM_001304718.1:c.-71A>G
NM_000314.7:c.521A>G
NM_001304717.5:c.1040A>G
NM_001304718.2:c.-71A>G

Likely Pathogenic

Met criteria codes 5

PS2 PP2 PP3 PM2_Supporting PS4_Supporting

Not Met criteria codes 1

BP4

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 3.0.0*
Criteria Specification Approval History
Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

NM_000314.8(PTEN):c.521A>G (p.Tyr174Cys) variant 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 3.0.0). 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. (Internal laboratory contributor(s) SCV000602123.1) PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (Internal laboratory contributor(s) SCV000602123.1). PM2_P: Absent in large sequenced populations (PMID 27535533). 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=0.978).

Met criteria codes

PS2	✔	De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (Internal laboratory contributor(s) SCV000602123.1)
PP2	✔	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=0.978)
PM2_Supporting	✔	PM2_Supporting: Absent in large sequenced populations in the gnomAD cohort. (PMID 27535533).
PS4_Supporting	✔	PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (Internal laboratory contributor(s) SCV000602123.1).

Not Met criteria codes

BP4	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

Showing 1 to 1 of 1 rows

The information on this website is not intended for direct diagnostic use or medical decision-making without review by a genetics professional. Individuals should not change their health behavior solely on the basis of information contained on this website. If you have questions about the information contained on this website, please see a health care professional.

ClinGen Terms of Use.
¤ Powered by BCM's Genboree.