

Variant: NM_000314.6(PTEN):c.517C>T (p.Arg173Cys)

CA000498 [↗](#)

189500 (ClinVar) [↗](#)

Gene: PTEN (HGNC:5728)

Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)

Inheritance Mode: Autosomal dominant inheritance

UID: 8f2c1cdc-0442-4dc1-ace0-ee811465348e

Approved on: 2017-10-18

Published on: 2018-12-10

HGVS expressions

NM_000314.6:c.517C>T

NM_000314.6(PTEN):c.517C>T (p.Arg173Cys)

NC_000010.11:g.87952142C>T

CM000672.2:g.87952142C>T

NC_000010.10:g.89711899C>T

CM000672.1:g.89711899C>T

NC_000010.9:g.89701879C>T

NG_007466.2:g.93704C>T

NM_000314.5:c.517C>T

NM_001304717.2:c.1036C>T

NM_001304718.1:c.-75C>T

NM_000314.7:c.517C>T

NM_001304717.5:c.1036C>T

NM_001304718.2:c.-75C>T

ENST00000371953.7:c.517C>T

Pathogenic

Met criteria codes **6**

PS2_Very Strong PS3 PP1 PP2

PM2 PS4_Moderate

Evidence Links **0**

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.517C>T (p.R173C) meets criteria to be classified as pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (Mester et al. 2018; manuscript in preparation). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PS2_VS: At least two proven OR one proven plus two assumed de novo observations in a patient with the disease and no family history. (PMID 17526800, PMID 22628360, internal laboratory contributor(s) SCV000222220.10) PS3: Phosphatase activity <50% of wild type. (PMID 10866302) PM2: Absent in large sequenced populations (PMID 27535533). PS4_M: Probands with phenotype specificity score of 2-3.5. (PMID 22628360, PMID 17526800, PMID 17526801) PP1: Co-segregation with disease in multiple affected family members, with 3 or 4 meioses observed. (PMID 22628360) 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.

Met criteria codes

PS2_Very Strong	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP2	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4_Moderate	✓	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.