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_000314.6(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
UUID: 8f2c1cdc-0442-4dc1-ace0-ee811465348e
Approved on: 2017-10-18
Published on: 2018-12-10

HGVS expressions

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

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