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.510T>A (p.Ser170Arg)

CA000492

7815 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 67ade3e6-3038-4022-903b-9caf929577ba
Approved on: 2018-04-06
Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.510T>A (p.Ser170Arg)
NM_000314.6:c.510T>A
NC_000010.11:g.87952135T>A
CM000672.2:g.87952135T>A
NC_000010.10:g.89711892T>A
CM000672.1:g.89711892T>A
NC_000010.9:g.89701872T>A
NG_007466.2:g.93697T>A
ENST00000371953.8:c.510T>A
ENST00000371953.7:c.510T>A
NM_000314.5:c.510T>A
NM_001304717.2:c.1029T>A
NM_001304718.1:c.-82T>A
NM_000314.7:c.510T>A
NM_001304717.5:c.1029T>A
NM_001304718.2:c.-82T>A
NM_000314.8:c.510T>A
NM_000314.8(PTEN):c.510T>A (p.Ser170Arg)

Pathogenic

Met criteria codes 5

PP2 PP1 PM2 PS3 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.510T>A (p.Ser170Arg) 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). PS3: Phosphatase activity <50% of wild type (PMID 17942903, PMID 10866302, PMID 9256433, PMID 21828076 PS4_M: Probands with phenotype specificity score of 2-3.5. (PMID 17526800, PMID 23335809, PMID 30528446) PM2: Absent in large sequenced populations (PMID 27535533) PP1: Co-segregation with disease in multiple affected

family members, with 3 or 4 meioses observed. (PMID 17526800, PMID 9241266) 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

PP2	✔	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
PM2	✔	Absent gnomAD
PS3	✔	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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