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Variant: NM_000314.6(PTEN):c.892C>T (p.Gln298Ter)

CA000219

187657 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: f890d81d-90ed-4411-a0fd-a89669dd616a
Approved on: 2017-10-18
Published on: 2018-12-10

HGVS expressions

NM_000314.6:c.892C>T
NM_000314.6(PTEN):c.892C>T (p.Gln298Ter)
NC_000010.11:g.87960984C>T
CM000672.2:g.87960984C>T
NC_000010.10:g.89720741C>T
CM000672.1:g.89720741C>T
NC_000010.9:g.89710721C>T
NG_007466.2:g.102546C>T
NM_000314.5:c.892C>T
NM_001304717.2:c.1411C>T
NM_001304718.1:c.301C>T
NM_000314.7:c.892C>T
NM_001304717.5:c.1411C>T
NM_001304718.2:c.301C>T
ENST00000371953.7:c.892C>T
ENST00000472832.2:n.319C>T

Pathogenic

Met criteria codes 3

PM2 PS4_Supporting PVS1

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.892C>T (p.Q298X) 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). PVS1: Null variant predicted to result in nonsense-mediated decay or causing truncation/frameshift at or 5’ to c.1121 (NM_000314.4). PM2: Absent in large sequenced populations (PMID 27535533). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID 23470840)

Met criteria codes

PM2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4_Supporting	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PVS1	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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