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Variant: NM_000314.6(PTEN):c.469G>T (p.Glu157Ter)

CA000467

7814 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: c8a8caba-2251-491c-8758-14da15b51065
Approved on: 2018-10-30
Published on: 2018-12-10

HGVS expressions

NM_000314.6:c.469G>T
NM_000314.6(PTEN):c.469G>T (p.Glu157Ter)
NC_000010.11:g.87933228G>T
CM000672.2:g.87933228G>T
NC_000010.10:g.89692985G>T
CM000672.1:g.89692985G>T
NC_000010.9:g.89682965G>T
NG_007466.2:g.74790G>T
NM_000314.5:c.469G>T
NM_001304717.2:c.988G>T
NM_001304718.1:c.-282G>T
NM_000314.7:c.469G>T
NM_001304717.5:c.988G>T
NM_001304718.2:c.-282G>T
ENST00000371953.7:c.469G>T
ENST00000498703.1:n.295G>T
ENST00000610634.1:c.367G>T

Pathogenic

Met criteria codes 3

PVS1 PM2 PS4_Supporting

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.469G>T (p.E157X) 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). 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 8071972)

Met criteria codes

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

Curation History

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