

Variant: NM_000314.6(PTEN):c.737C>T (p.Pro246Leu)

CA000559 [↗](#)

142269 (ClinVar) [↗](#)

Gene: PTEN ([HGNC:5728](#))

Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))

Inheritance Mode: Autosomal dominant inheritance

UUID: 835a14e6-4a41-47df-a1ff-271c872adf39

Approved on: 2017-10-18

Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.737C>T (p.Pro246Leu)

NM_000314.6:c.737C>T

NM_000314.5:c.737C>T

NM_001304717.2:c.1256C>T

NM_001304718.1:c.146C>T

NM_000314.7:c.737C>T

NM_001304717.5:c.1256C>T

NM_001304718.2:c.146C>T

ENST00000371953.7:c.737C>T

ENST00000472832.2:n.164C>T

NC_000010.11:g.87957955C>T

CM000672.2:g.87957955C>T

NC_000010.10:g.89717712C>T

CM000672.1:g.89717712C>T

NC_000010.9:g.89707692C>T

NG_007466.2:g.99517C>T

Pathogenic

Met criteria codes **4**

PS2_Very Strong

PS4

PP2

PM2

Evidence Links **0**

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) **Criteria Specifications for this VCEP**

Evidence submitted by expert panel

PTEN VCEP

PTEN c.737C>T (p.P246L) 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 23934111, internal laboratory contributor(s) SCV000222227.10) PS4: Probands with phenotype specificity score of 4-15.5. (PMID 24375884, PMID 10076877, PMID 22261759, PMID 23934111, internal laboratory contributor(s) SCV000222227.10) PM2: Absent in large sequenced populations. (PMID 27535533). 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
PS4			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

Curation History



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