

Variant: NM_000314.8(PTEN):c.542T>C (p.Leu181Pro)

Version: 1.0

CA377484387 [↗](#)

825730 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 0c0487d6-9459-4255-b69d-51bb10a6f8a9

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HGVS expressions

NM_000314.8:c.542T>C

NM_000314.8(PTEN):c.542T>C (p.Leu181Pro)

NC_000010.11:g.87952167T>C

CM000672.2:g.87952167T>C

NC_000010.10:g.89711924T>C

CM000672.1:g.89711924T>C

NC_000010.9:g.89701904T>C

NG_007466.2:g.93729T>C

ENST00000700029.2:c.542T>C

ENST00000710265.1:c.542T>C

ENST00000472832.3:c.542T>C

ENST00000688158.2:n.1277T>C

ENST00000688922.2:c.*372T>C

ENST00000700021.1:c.497T>C

ENST00000700022.1:c.493-5686T>C

ENST00000700023.1:n.1700T>C

ENST00000700024.1:n.1934T>C

ENST00000700025.1:n.1311T>C

ENST00000700029.1:c.376T>C

ENST00000706954.1:c.542T>C

ENST00000706955.1:c.*577T>C

ENST00000686459.1:c.*128T>C

ENST00000688158.1:c.*653T>C

ENST00000688308.1:c.542T>C

ENST00000688922.1:c.463T>C

ENST00000693560.1:c.1061T>C

ENST00000371953.8:c.542T>C

ENST00000371953.7:c.542T>C

NM_000314.5:c.542T>C

NM_000314.6:c.542T>C

NM_001304717.2:c.1061T>C

NM_001304718.1:c.-50T>C

NM_000314.7:c.542T>C

NM_001304717.5:c.1061T>C

NM_001304718.2:c.-50T>C

Pathogenic

Met criteria codes 5

PS4_SupportingPS2PS3PP2PM2

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specification: ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines Version 2

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.542T>C (p.Leu181Pro) 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). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor(s) SCV001524663.1) PS3: Phosphatase activity <50% (PMID 29706350, PMID 21828076) 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. PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID 17526801, internal laboratory contributor(s) SCV000579246.1)

Met criteria codes

PS4_Supporting	i ✓	Peds score of 4 = ½ proband point (Tan et al. 2007 PMID: 17526801) and CC score of 29 = ½ proband point (SCV000579246.1).
PS2	i ✓	Identified in a male proband in teens with thoracic aortic aneurysm, abnormal tricuspid valve morphology, abnormal aortic arch morphology, myopia, macrocephaly, cognitive impairment, and autistic behavior. It was de novo by trio exome sequencing (SCV001524663.1).
PS3	i ✓	>50% reduction in phosphatase activity (PMID: 29706350 and PMID: 21828076)
PP2	i ✓	missense constraint
PM2	✓	Absent in gnomAD.

Curation History



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