

Variant: *NM_000314.7(PTEN):c.42G>A (p.Arg14=)*

Version: 1.1

CA16619040 [🔗](#)

420491 (ClinVar) [🔗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 9cbc440e-91aa-45a2-bf57-6c217f5bdb7

Approved on: 2021/6/4

Published on: 2022/9/30

HGVS expressions

NM_000314.7:c.42G>A

NM_000314.7(PTEN):c.42G>A (p.Arg14=)

NC_000010.11:g.87864511G>A

CM000672.2:g.87864511G>A

NC_000010.10:g.89624268G>A

CM000672.1:g.89624268G>A

NC_000010.9:g.89614248G>A

NG_007466.2:g.6073G>A

NG_033079.1:g.3927C>T

ENST00000700029.2:c.42G>A

ENST00000710265.1:c.42G>A

ENST00000472832.3:c.42G>A

ENST00000688922.2:c.42G>A

ENST00000700021.1:c.42G>A

ENST00000700022.1:c.42G>A

ENST00000706954.1:c.42G>A

ENST00000706955.1:c.42G>A

ENST00000686459.1:c.42G>A

ENST00000688158.1:c.42G>A

ENST00000688308.1:c.42G>A

ENST00000693560.1:c.561G>A

ENST00000371953.8:c.42G>A

ENST00000371953.7:c.42G>A

ENST00000462694.1:n.44G>A

ENST00000487939.1:n.63G>A

ENST00000610634.1:c.-61G>A

ENST00000618586.1:n.11G>A

NM_000314.5:c.42G>A

NM_000314.6:c.42G>A

NM_001304717.2:c.561G>A

NM_001304718.1:c.-664G>A

NM_001304717.5:c.561G>A

NM_001304718.2:c.-664G>A

NM_000314.8:c.42G>A

Uncertain Significance

Met criteria codes 1

PM2

Not Met criteria codes 3

BP5BP7BP4

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.42G>A (p.Arg14=) is currently classified as a variant of uncertain significance 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). PM2: Absent in large sequenced populations (PMID 27535533).

Met criteria codes

PM2

✓

Absent in gnomAD

Not Met criteria codes

BP5

i

✗

GDX1: Middle Eastern F child with features of BBS (obesity, night blindness, mild ID, polydactyly, heart defect) and HOMOZYGOUS PATH BBS7 variant – may be able to count towards BP5 when more co-occurrences become available. Father POSITIVE for PTEN variant, healthy, large family, no cancers/other hx concerning for PTEN.

BP7

i

✗

Synonymous. PhyloP score is 5.94; nucleotide is highly conserved.

BP4

i

✗

Intron 1 SDS not predicted by in silico tools

Curation History

▾

▾

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