

Variant: NM_000314.6(PTEN):c.914G>A (p.Ser305Asn)

Version: 1.0

CA000632 [↗](#)

127696 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 59aacee3-4abf-400b-9713-4c28e3258b75

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HGVS expressions

NM_000314.6:c.914G>A

NM_000314.6(PTEN):c.914G>A (p.Ser305Asn)

NC_000010.11:g.87961006G>A

CM000672.2:g.87961006G>A

NC_000010.10:g.89720763G>A

CM000672.1:g.89720763G>A

NC_000010.9:g.89710743G>A

NG_007466.2:g.102568G>A

ENST00000700029.2:c.1007G>A

ENST00000710265.1:c.914G>A

ENST00000472832.3:c.914G>A

ENST00000688158.2:n.1649G>A

ENST00000688922.2:c.*744G>A

ENST00000700021.1:c.869G>A

ENST00000700022.1:c.*253G>A

ENST00000700023.1:n.2072G>A

ENST00000700024.1:n.2306G>A

ENST00000700025.1:n.1683G>A

ENST00000700026.1:n.551G>A

ENST00000706954.1:c.914G>A

ENST00000706955.1:c.*949G>A

ENST00000686459.1:c.*500G>A

ENST00000688158.1:c.*1025G>A

ENST00000688308.1:c.914G>A

ENST00000688922.1:c.835G>A

ENST00000693560.1:c.1433G>A

ENST00000371953.8:c.914G>A

ENST00000371953.7:c.914G>A

ENST00000472832.2:c.341G>A

NM_000314.5:c.914G>A

NM_001304717.2:c.1433G>A

NM_001304718.1:c.323G>A

NM_000314.7:c.914G>A

NM_001304717.5:c.1433G>A

NM_001304718.2:c.323G>A

NM_000314.8:c.914G>A

Uncertain Significance

Met criteria codes 3

PM2 BS3_Supporting PP2

Not Met criteria codes 22

PM1 PM3 PM5 PM4 PM6
BA1 BS2 BS1 BS4 BP4 BP3
BP1 BP2 BP5 BP7 PS1 PS2
PS3 PS4 PVS1 PP1 PP3

Evidence Links 1

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information **!**

[↗](#) Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.914G>A (p.S305N) 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).** **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.** **BS3_P: In vitro or in vivo functional study or studies showing no damaging effect on protein function but BS3 not met. (PMID 29706350)**

Met criteria codes

PM2	✓	Absent gnomAD
BS3_Supporting	✓	Mighell 2018 PMID 29706350 High-throughput phosphatase assay results in WT-like range (-0.76). PubMed:29706350 ↗
PP2	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Not Met criteria codes

PM1	✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM3	✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5	✗	ClinVar entry for c.915T>G (p.Ser305Arg) [ID: 230040]. 1 submitter who reports 1 individual with a hereditary cancer-predisposing syndrome phenotype. Variant not found in the literature.
PM4	✗	

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

PM6	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP3	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP1	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP2	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP5	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP7	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS2	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	✘	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
PVS1	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP3	✘	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History [↗](#)

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