The ClinGen Evidence Repository is an **FDA-recognized human genetic variant database** containing expert-curated assertions regarding variants' pathogenicity and supporting evidence summaries. [Disclaimer]

Variant: NM_000314.7(PTEN):c.319G>T (p.Asp107Tyr)

Version: 1.0

CA377482178

484614 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: d559324d-7f4d-426b-8591-b4f7f58350c2
Approved on: 2021/10/29
Published on: 2021/10/29

HGVS expressions

NM_000314.7:c.319G>T
NM_000314.7(PTEN):c.319G>T (p.Asp107Tyr)
NC_000010.11:g.87933078G>T
CM000672.2:g.87933078G>T
NC_000010.10:g.89692835G>T
CM000672.1:g.89692835G>T
NC_000010.9:g.89682815G>T
NG_007466.2:g.74640G>T
ENST00000700029.2:c.319G>T
ENST00000710265.1:c.319G>T
ENST00000472832.3:c.319G>T
ENST00000688158.2:n.1054G>T
ENST00000688922.2:c.*149G>T
ENST00000700021.1:c.274G>T
ENST00000700022.1:c.319G>T
ENST00000700029.1:c.153G>T
ENST00000706954.1:c.319G>T
ENST00000706955.1:c.*354G>T
ENST00000686459.1:c.319G>T
ENST00000688158.1:c.*430G>T
ENST00000688308.1:c.319G>T
ENST00000688922.1:c.240G>T
ENST00000693560.1:c.838G>T
ENST00000371953.8:c.319G>T
ENST00000371953.7:c.319G>T
ENST00000498703.1:n.145G>T
ENST00000610634.1:c.217G>T
NM_000314.5:c.319G>T
NM_000314.6:c.319G>T
NM_001304717.2:c.838G>T
NM_001304718.1:c.-432G>T
NM_001304717.5:c.838G>T
NM_001304718.2:c.-432G>T
NM_000314.8:c.319G>T

Pathogenic

Met criteria codes 5

PS3 PP2 PM5 PM2 PS4_Supporting

Not Met criteria codes 3

PP1 BA1 BS1

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.319G>T (p.Asp107Tyr) 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). PS3: Phosphatase activity <50% of wild type (PMID 10866302, PMID 29706350, PMID 29785012) PM2: Absent in large sequenced populations (PMID 27535533) PM5: Missense change at an amino acid residue where a different missense change determined to be pathogenic or likely pathogenic and with equal or lesser BLOSUM62 score has been seen before (ClinVar Variation ID 372481, SCV000930124) 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 23382303)

Met criteria codes

PS3	✔	Han 2000 PMID 10866302, Mighell 2018 PMID 29706350, Matreyek 2018 PMID 29785012
PP2	✔	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5	✔	D107V pathogenic and reviewed by PTEN expert panel and same score (-3) for BLOSUM62
PM2	✔	absent in gnomAD
PS4_Supporting	✔	Granados 2013 PMID 23382303; 1 point

Not Met criteria codes

PP1	✖	Granados 2013 PMID 23382303; 1 meiosis
BA1	✖	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

Curation History

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