

Variant: NM_000314.8(PTEN):c.39_41del (p.Arg15del)

Version: 1.0

CA645369492 [↗](#)

428199 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 733edbab-4281-44cd-b894-8ec48422fab7

Approved on: 2021/6/4

Published on: 2022/9/30

HGVS expressions

NM_000314.8(PTEN):c.39_41del (p.Arg15del)

NM_000314.8:c.39_41del

NC_000010.11:g.87864508_87864510del

CM000672.2:g.87864508_87864510del

NC_000010.10:g.89624265_89624267del

CM000672.1:g.89624265_89624267del

NC_000010.9:g.89614245_89614247del

NG_007466.2:g.6070_6072del

NG_033079.1:g.3928_3930del

ENST00000700029.2:c.39_41del

ENST00000710265.1:c.39_41del

ENST00000472832.3:c.39_41del

ENST00000688922.2:c.39_41del

ENST00000700021.1:c.39_41del

ENST00000700022.1:c.39_41del

ENST00000706954.1:c.39_41del

ENST00000706955.1:c.39_41del

ENST00000686459.1:c.39_41del

ENST00000688158.1:c.39_41del

ENST00000688308.1:c.39_41del

ENST00000693560.1:c.558_560del

ENST00000371953.8:c.39_41del

ENST00000371953.7:c.39_41del

ENST00000462694.1:n.41_43del

ENST00000487939.1:n.60_62del

ENST00000610634.1:c.-64_-62del

ENST00000618586.1:n.8_10del

NM_000314.5:c.39_41del

NM_000314.6:c.39_41del

NM_001304717.2:c.558_560del

NM_001304718.1:c.-667_-665del

NM_000314.7:c.39_41del

NM_001304717.5:c.558_560del

NM_001304718.2:c.-667_-665del

Uncertain Significance

Met criteria codes 2

PM2PS4_Supporting

Not Met criteria codes 1

PS3

Evidence Links 1

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.39_41del (p.Arg15del) 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). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID 30528446)

Met criteria codes

PM2

✓

Absent in gnomAD

PS4_Supporting

i

✓

4y11m old proband (age at evaluation was 2y4m) with DD/ID, ASD, brain MRI anomalies, vascular malformation (loop of the vertebral artery), HC of 57 cm (+7.3 SD). Peds score of 7 = 1 proband point (PMID: 30528446)

Not Met criteria codes

PS3

i

✗

Hypomorphic range -1.22939927 (TRUE) in Mighell et al. 2018 (PMID: 29706350)

Humanized yeast model of lipid phosphatase activity in vivo. This variant was in the hypomorphic range -1.22939927 (TRUE), so PS3 cannot be applied. [PubMed:29706350](#)

Curation History

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