

Variant: NM_000314.8(PTEN):c.270dup (p.Glu91Ter)

CA645369474 [↗](#)

428271 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a3be49ba-5c93-4f8f-a2a8-e7d30a583ad9

Approved on: 2018-11-28

Published on: 2018-12-10

HGVS expressions

NM_000314.8(PTEN):c.270dup (p.Glu91Ter)

NM_000314.8:c.270dup

NC_000010.11:g.87933029dup

CM000672.2:g.87933029dup

NC_000010.10:g.89692786dup

CM000672.1:g.89692786dup

NC_000010.9:g.89682766dup

NG_007466.2:g.74591dup

ENST00000686459.1:c.270dup

ENST00000688158.1:c.*381dup

ENST00000688308.1:c.270dup

ENST00000688922.1:c.191dup

ENST00000693560.1:c.789dup

ENST00000371953.8:c.270dup

ENST00000371953.7:c.270dup

ENST00000498703.1:n.96dup

ENST00000610634.1:c.168dup

NM_000314.5:c.270dup

NM_000314.6:c.270dup

NM_001304717.2:c.789dup

NM_001304718.1:c.-481dup

NM_000314.7:c.270dup

NM_001304717.5:c.789dup

NM_001304718.2:c.-481dup

Likely Pathogenic

Met criteria codes 2

PM2_Supporting

PVS1

Not Met criteria codes 24

BP5

BP7

BP2

BP3

BP1

BP4

BS2

BS4

BS3

BS1

PP4

PP1

PP2

PP3

PM6

PM5

PM3

PM1

PM4

PS1

PS2

PS4

PS3

BA1

Evidence Links 0

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information

[↗](#) **Criteria Specification:** ClinGen PTEN Expert Panel
Specifications to the ACMG/AMP Variant Interpretation
Guidelines for PTEN Version 3.0.0

[↗](#) **Criteria Specification Approval History**

[↗](#) **Criteria Specifications for this VCEP**

PTEN VCEP

NM_000314.8(PTEN):c.270dup (p.Glu91Ter) variant meets criteria to be classified as likely pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.0.0). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PVS1: Null variant predicted to result in nonsense-mediated decay or causing truncation/frameshift at or 5’ to c.1121 (NM_000314.8). PM2_P: Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort (PMID:27535533).

Met criteria codes

PM2_Supporting			Absent in large sequenced populations OR present at extremely low (<0.00001, 0.001%) allele frequency in the gnomAD cohort (PMID:27535533).
PVS1			PVS1: Null variant predicted to result in nonsense-mediated decay or causing truncation/frameshift at or 5’ to c.1121 (NM_000314.8).

Not Met criteria codes

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
BP2			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
BP4			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
BS4			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline			
BS3			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1			Absent in gnomAD
PP4			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
PP2			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
PM6			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM5			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
PM1			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM4			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
PS4			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

BA1



Absent in gnomAD

Curation History

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