

Variant: NM_000314.7(PTEN):c.1133_1136del (p.Arg378fs)

Version: 1.1

CA16619071 [↗](#)
421055 (ClinVar) [↗](#)
Gene: PTEN ([HGNC:5728](#))
Condition: PTEN hamartoma tumor syndrome ([MONDO:0017623](#))
Inheritance Mode: Autosomal dominant inheritance
UUID: fd86daf3-07ce-4482-918a-b7dd2d40fd3b
Approved on: 2024/2/9
Published on: 2024/3/4

HGVS expressions

NM_000314.7(PTEN):c.1133_1136del (p.Arg378fs)
NM_000314.7:c.1133_1136del
NC_000010.11:g.87965393_87965396del
CM000672.2:g.87965393_87965396del
NC_000010.10:g.89725150_89725153del
CM000672.1:g.89725150_89725153del
NC_000010.9:g.89715130_89715133del
NG_007466.2:g.106955_106958del
ENST00000700029.2:c.1226_1229del
ENST00000710265.1:c.*162_*165del
ENST00000688158.2:n.1868_1871del
ENST00000688922.2:c.*963_*966del
ENST00000700021.1:c.1088_1091del
ENST00000700022.1:c.*472_*475del
ENST00000700023.1:n.2291_2294del
ENST00000700024.1:n.2525_2528del
ENST00000706954.1:c.1133_1136del
ENST00000706955.1:c.*1168_*1171del
ENST00000686459.1:c.*719_*722del
ENST00000688158.1:c.*1244_*1247del
ENST00000688308.1:c.1133_1136del
ENST00000688922.1:c.1054_1057del
ENST00000693560.1:c.1652_1655del
ENST00000371953.8:c.1133_1136del
ENST00000371953.7:c.1133_1136del
NM_000314.5:c.1133_1136del
NM_000314.6:c.1133_1136del
NM_001304717.2:c.1652_1655del
NM_001304718.1:c.542_545del
NM_001304717.5:c.1652_1655del
NM_001304718.2:c.542_545del
NM_000314.8:c.1133_1136del

Uncertain Significance

Met criteria codes2

PM2_SupportingPM4

Not Met criteria codes23

BA1BS1BS4BS3BS2BP5BP7BP4BP3BP1BP2PVS1PS1PS2PS3PP1PP2PP3PP4PM6PM1PM3PM5

Evidence Links0

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

Evidence submitted by expert panel

PTEN VCEP			
NM_000314.8(PTEN):c.1133_1136del (p.Arg378IlefsTer37) is currently classified as a variant of uncertain significance 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). PM2_P: Absent in large sequenced populations (PMID 27535533). PM4: Variant causes protein extension (NM_000314.4)			
Met criteria codes			
PM2_Supporting		✓	absent gnomAD
PM4		✓	Variant causes protein extension
Not Met criteria codes			
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
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
BS2		✗	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
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
PVS1			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
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
PP4			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

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

Curation History 		
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