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_000314.6(PTEN):c.103A>G (p.Met35Val)

CA10578906

231916 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 4f6345e3-25b1-48cd-9697-1a5624715071
Approved on: 2017-10-18
Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.103A>G (p.Met35Val)
NM_000314.6:c.103A>G
NC_000010.11:g.87894048A>G
CM000672.2:g.87894048A>G
NC_000010.10:g.89653805A>G
CM000672.1:g.89653805A>G
NC_000010.9:g.89643785A>G
NG_007466.2:g.35610A>G
ENST00000700029.2:c.103A>G
ENST00000710265.1:c.103A>G
ENST00000472832.3:c.103A>G
ENST00000688158.2:n.899+13610A>G
ENST00000688922.2:c.103A>G
ENST00000700021.1:c.103A>G
ENST00000700022.1:c.103A>G
ENST00000706954.1:c.103A>G
ENST00000706955.1:c.*138A>G
ENST00000686459.1:c.103A>G
ENST00000688158.1:c.*275+13610A>G
ENST00000688308.1:c.103A>G
ENST00000693560.1:c.622A>G
ENST00000371953.8:c.103A>G
ENST00000371953.7:c.103A>G
ENST00000462694.1:n.105A>G
ENST00000610634.1:c.1A>G
NM_000314.5:c.103A>G
NM_001304717.2:c.622A>G
NM_001304718.1:c.-603A>G
NM_000314.7:c.103A>G
NM_001304717.5:c.622A>G
NM_001304718.2:c.-603A>G
NM_000314.8:c.103A>G
NM_000314.8(PTEN):c.103A>G (p.Met35Val)

Met criteria codes 6

Expert Panel

Pathogenic

PP2 PP3 PS2_Very Strong PM2_Supporting PS4_Supporting PS3_Supporting

Not Met criteria codes 20

PS1 PP1 PP4 PM5 PM1 PM4 PM3 PM6 BA1 BS4 BS3 BS1 BS2 PVS1 BP7 BP5 BP3 BP2 BP1 BP4

Evidence Links 0

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.103A>G (p.Met35Val) meets criteria to be classified as 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). PS2_VS: At least two proven de novo observations in a patient with the disease and no family history. (internal laboratory contributors: SCV000275912.7, SCV000565444.8). PS3_P: Abnormal in vitro cellular assay or transgenic model with phenotype different from wild type that does not meet PS3 (Post et al. 2020 PMID: 32350270: pAKT levels similar to C124S). PS4_P: Proband(s) with phenotype specificity score of 1-1.5. (PMID: 27531073, internal laboratory contributor: SCV000275912.7). PM2_P: 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. PP3: REVEL score > 0.7 (score of this variant =0.953)

Met criteria codes

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.
PP3	✔	REVEL score > 0.7 (score of this variant =0.953)
PS2_Very Strong	✔	At least two proven de novo observations in a patient with the disease and no family history. (internal laboratory contributors: SCV000275912.7, SCV000565444.8)
PM2_Supporting	✔	Absent in gnomAD.
PS4_Supporting	✔	Proband(s) with phenotype specificity score of 1-1.5. (PMID: 27531073, internal laboratory contributor: SCV000275912.7)
PS3_Supporting	✔	Abnormal in vitro cellular assay or transgenic model with phenotype different from wild type that does not meet PS3 (Post et al. 2020 PMID: 32350270: pAKT levels similar to C124S).

Not Met criteria codes

PS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PP1		✖	2 meioses in internal lab cases per March 2020 meeting - check for more in future cases.
PP4		✖	Wrapped into PS4
PM5		✖	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
PM3		✖	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
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		✖	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
PVS1		✖	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
BP5		✖	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
BP2		✖	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

Curation History

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