

Variant: NM_000314.6(PTEN):c.407G>A (p.Cys136Tyr)

CA000451 [↗](#)

189406 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: 11de4e4a-dc77-4323-92c5-151d4868419d

Approved on: 2017-10-18

Published on: 2018-12-10

HGVS expressions

NM_000314.6(PTEN):c.407G>A (p.Cys136Tyr)

NM_000314.6:c.407G>A

NC_000010.11:g.87933166G>A

CM000672.2:g.87933166G>A

NC_000010.10:g.89692923G>A

CM000672.1:g.89692923G>A

NC_000010.9:g.89682903G>A

NG_007466.2:g.74728G>A

NM_000314.5:c.407G>A

NM_001304717.2:c.926G>A

NM_001304718.1:c.-344G>A

NM_000314.7:c.407G>A

NM_001304717.5:c.926G>A

NM_001304718.2:c.-344G>A

ENST00000371953.7:c.407G>A

ENST00000498703.1:n.233G>A

ENST00000610634.1:c.305G>A

Pathogenic

Met criteria codes **5**

PM2

PS2

PS3

PP2

PS4_Supporting

Evidence Links **0**

Expert Panel

PTEN VCEP [↗](#)

Criteria Specification Information **!**

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

Evidence submitted by expert panel

PTEN VCEP

PTEN c.407G>A (p.C136Y) meets criteria to be classified as pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (Mester et al. 2018; manuscript in preparation). Please see a summary of the rules and criteria codes in the “PTEN ACMG Specifications Summary” document (assertion method column). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (Internal laboratory contributor(s) SCV000222113.9) PS3: Phosphatase activity <50% of wild-type OR RNA, mini-gene, or other assay shows impact on splicing. (PMID 10866302) PM2: 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. PS4_P: Probands with specificity score of 1-1.5. (PMID 9735393, internal laboratory contributor(s) SCV000222113.9)

Met criteria codes			
PM2		✓	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
PP2		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS4_Supporting		✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

Curation History

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