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_000314.7(PTEN):c.830C>G (p.Thr277Arg)

Version: 1.0

CA377485523

428268 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 579af9bf-2984-45d2-8e0a-bd0802a7860b
Approved on: 2020/3/23
Published on: 2020/3/26

HGVS expressions

NM_000314.7:c.830C>G
NM_000314.7(PTEN):c.830C>G (p.Thr277Arg)
NC_000010.11:g.87960922C>G
CM000672.2:g.87960922C>G
NC_000010.10:g.89720679C>G
CM000672.1:g.89720679C>G
NC_000010.9:g.89710659C>G
NG_007466.2:g.102484C>G
ENST00000700029.2:c.923C>G
ENST00000710265.1:c.830C>G
ENST00000472832.3:c.830C>G
ENST00000688158.2:n.1565C>G
ENST00000688922.2:c.*660C>G
ENST00000700021.1:c.785C>G
ENST00000700022.1:c.*169C>G
ENST00000700023.1:n.1988C>G
ENST00000700024.1:n.2222C>G
ENST00000700025.1:n.1599C>G
ENST00000700026.1:n.467C>G
ENST00000700029.1:c.757C>G
ENST00000706954.1:c.830C>G
ENST00000706955.1:c.*865C>G
ENST00000686459.1:c.*416C>G
ENST00000688158.1:c.*941C>G
ENST00000688308.1:c.830C>G
ENST00000688922.1:c.751C>G
ENST00000693560.1:c.1349C>G
ENST00000371953.8:c.830C>G
ENST00000371953.7:c.830C>G
ENST00000472832.2:c.257C>G
NM_000314.5:c.830C>G
NM_000314.6:c.830C>G
NM_001304717.2:c.1349C>G
NM_001304718.1:c.239C>G
NM_001304717.5:c.1349C>G
NM_001304718.2:c.239C>G
NM_000314.8:c.830C>G

Likely Pathogenic

Met criteria codes 4

PP1_Moderate PM2 PS3 PP2

Not Met criteria codes 22

PVS1 BS2 BS1 BS4 BS3
BP5 BP7 BP4 BP3 BP1 BP2
PM6 PM1 PM3 PM5 PM4
PS1 PS2 PS4 BA1 PP3 PP4

Evidence Links 3

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.830C>G (p.Thr277Arg) meets criteria to be classified as likely pathogenic 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). PS3: Phosphatase activity <50% of wild type (PMID 29706350) PM2: Absent in large sequenced populations PP1_M: Co-segregation with disease in multiple affected family members, with 5 or 6 meioses observed. (PMID 23335809) 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.

Met criteria codes

PP1_Moderate	✔	Pedigree provided by Dr. Longy confirming 5 meioses. PubMed:23335809
PM2	✔	Not present in gnomAD (coverage >30x).
PS3	✔	Mighell et al. 2018 High-throughput functional assay testing PTEN lipid phosphatase activity. T277R had a score of -3.839319668 and was 'TRUE', which means this variant had truncation-like function. High-throughput functional assay testing PTEN lipid phosphatase activity. T277R had a score of -3.839319668 and was 'TRUE', which means this variant had truncation-like function. PubMed:29706350
PP2	✔	Missense

Not Met criteria codes

PVS1	✖	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
BS1		✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4		✖	PubMed:23335809
BS3		✖	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
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
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
BA1		✖	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		✖	PubMed:20712882

Curation History

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