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.1212A>T (p.Ter404Cys)

Version: 1.0

CA10578936

234144 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: fc24cd5e-624d-4b84-b652-6f843664f53b
Approved on: 2019/11/22
Published on: 2019/12/11

HGVS expressions

NM_000314.7(PTEN):c.1212A>T (p.Ter404Cys)
NM_000314.7:c.1212A>T
NC_000010.11:g.87965472A>T
CM000672.2:g.87965472A>T
NC_000010.10:g.89725229A>T
CM000672.1:g.89725229A>T
NC_000010.9:g.89715209A>T
NG_007466.2:g.107034A>T
ENST00000700029.2:c.1305A>T
ENST00000710265.1:c.*241A>T
ENST00000688158.2:n.1947A>T
ENST00000688922.2:c.*1042A>T
ENST00000700021.1:c.1167A>T
ENST00000700022.1:c.*551A>T
ENST00000700023.1:n.2370A>T
ENST00000700024.1:n.2604A>T
ENST00000706954.1:c.1212A>T
ENST00000706955.1:c.*1247A>T
ENST00000686459.1:c.*798A>T
ENST00000688158.1:c.*1323A>T
ENST00000688308.1:c.1212A>T
ENST00000688922.1:c.1133A>T
ENST00000693560.1:c.1731A>T
ENST00000371953.8:c.1212A>T
ENST00000371953.7:c.1212A>T
NM_000314.5:c.1212A>T
NM_000314.6:c.1212A>T
NM_001304717.2:c.1731A>T
NM_001304718.1:c.621A>T
NM_001304717.5:c.1731A>T
NM_001304718.2:c.621A>T
NM_000314.8:c.1212A>T

Pathogenic

Met criteria codes 4

PM4 PM2 PS2 PS4_Moderate

Not Met criteria codes 21

PP1 PP2 PP3 PP4 PM1 PM3 PM5 BA1 PM6 BS2 BS1 BS4 BS3 PS1 PS3 BP4 BP3 BP1 BP2 BP5 BP7

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.1212A>T (p.Ter404CysextTer8) meets criteria to be classified as 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). PS2: De novo (both maternity and paternity confirmed) observation in a patient with the disease and no family history. (internal laboratory contributor ClinVar Organization ID: 26957) PM2: Absent in large sequenced populations (PMID 27535533). PM4: Variant causes protein extension. PS4_M: Probands with phenotype specificity score of 2-3.5. (internal laboratory contributors ClinVar Organization ID: 26957, SCV000278659.5)

Met criteria codes

PM4	✓	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM2	✓	Absent in gnomAD (coverage >40X at this position).
PS2	✓	Confirmed de novo via exome (PS2) in a male child with autism and OFC +5SD (1 point) (GeneDx).
PS4_Moderate	✓	Identified in a teenager with macrocephaly, thyroid cancer, and developmental delay (1 point) (Ambry Genetics). Confirmed de novo via exome (PS2) in a male child with autism and OFC +5SD (1 point) (GeneDx).

Not Met criteria codes

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		✖	Wrapped into PS4.
PM1	ℹ	✖	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	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BA1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PM6	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS2	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS4	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS1	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
PS3	ℹ	✖	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BP4	ℹ	✖	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	ℹ	✖	

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

Curation History

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