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Variant: NM_000314.7(PTEN):c.389G>C (p.Arg130Pro)

Version: 1.0

CA000439

142018 (ClinVar)

Gene: PTEN (HGNC:5728)
Condition: PTEN hamartoma tumor syndrome (MONDO:0017623)
Inheritance Mode: Autosomal dominant inheritance
UUID: 10bca2c6-405c-4f3a-919f-53db4e026879
Approved on: 2019/11/22
Published on: 2019/12/11

HGVS expressions

NM_000314.7:c.389G>C
NM_000314.7(PTEN):c.389G>C (p.Arg130Pro)
NC_000010.11:g.87933148G>C
CM000672.2:g.87933148G>C
NC_000010.10:g.89692905G>C
CM000672.1:g.89692905G>C
NC_000010.9:g.89682885G>C
NG_007466.2:g.74710G>C
ENST00000700029.2:c.389G>C
ENST00000710265.1:c.389G>C
ENST00000472832.3:c.389G>C
ENST00000688158.2:n.1124G>C
ENST00000688922.2:c.*219G>C
ENST00000700021.1:c.344G>C
ENST00000700022.1:c.389G>C
ENST00000700029.1:c.223G>C
ENST00000706954.1:c.389G>C
ENST00000706955.1:c.*424G>C
ENST00000686459.1:c.389G>C
ENST00000688158.1:c.*500G>C
ENST00000688308.1:c.389G>C
ENST00000688922.1:c.310G>C
ENST00000693560.1:c.908G>C
ENST00000371953.8:c.389G>C
ENST00000371953.7:c.389G>C
ENST00000498703.1:n.215G>C
ENST00000610634.1:c.287G>C
NM_000314.5:c.389G>C
NM_000314.6:c.389G>C
NM_001304717.2:c.908G>C
NM_001304718.1:c.-362G>C
NM_001304717.5:c.908G>C
NM_001304718.2:c.-362G>C
NM_000314.8:c.389G>C

Pathogenic

Met criteria codes 5

PS3 PP2 PM1 PM2 PS4_Supporting

Not Met criteria codes 19

PS1 PS2 PP1 PP3 PP4 PM3 PM4 PM6 BA1 BS1 BS4 BS3 BS2 BP5 BP7 BP4 BP3 BP1 BP2

Evidence Links 1

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.389G>C (p.Arg130Pro) 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). PS3: Phosphatase activity <50% of wild type (PMID 29706350) PM1: Located at a residue within a catalytic motif as defined by the ClinGen PTEN Expert Panel. 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: Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor SCV000185699.6)

Met criteria codes

PS3	✔	High-throughput functional assay testing PTEN Lipid Phosphatase Activity. R130P had a score of -3.153431338 and was 'true' indicating function was truncation-like for PS3.
		High-throughput functional assay testing PTEN Lipid Phosphatase Activity. R130P had a score of -3.153431338 and was 'true' indicating function was truncation-like for PS3. PubMed:29706350
PP2	✔	Missense.
PM1	✔	Affects amino acid position 130, which is in a catalytic motif.
PM2	✔	Not present in gnomAD (coverage >90X).
PS4_Supporting	✔	Proband in 60s with goiter, thyroid ca in 20s, kidney cancer in 60s, ganglioneuroma, >10 colon polyps, macrocephaly. (1 point) (Ambry Genetics).

Not Met criteria codes

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
PP1		✖	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. Proband in 60s with goiter, thyroid ca in 20s, kidney cancer in 60s, ganglioneuroma, >10 colon polyps, macrocephaly. (1 point) (Ambry Genetics).
PM3		✖	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
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
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

Curation History

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