

Variant: *NM_000314.7(PTEN):c.422A>C (p.His141Pro)*

Version: 1.0

CA337448 [↗](#)

216422 (ClinVar) [↗](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: c9d4dbc9-56ea-4ae3-a6b9-27fc09d9501c

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HGVS expressions

NM_000314.7(PTEN):c.422A>C (p.His141Pro)

NM_000314.7:c.422A>C

NC_000010.11:g.87933181A>C

CM000672.2:g.87933181A>C

NC_000010.10:g.89692938A>C

CM000672.1:g.89692938A>C

NC_000010.9:g.89682918A>C

NG_007466.2:g.74743A>C

ENST00000700029.2:c.422A>C

ENST00000710265.1:c.422A>C

ENST00000472832.3:c.422A>C

ENST00000688158.2:n.1157A>C

ENST00000688922.2:c.*252A>C

ENST00000700021.1:c.377A>C

ENST00000700022.1:c.422A>C

ENST00000700029.1:c.256A>C

ENST00000706954.1:c.422A>C

ENST00000706955.1:c.*457A>C

ENST00000686459.1:c.422A>C

ENST00000688158.1:c.*533A>C

ENST00000688308.1:c.422A>C

ENST00000688922.1:c.343A>C

ENST00000693560.1:c.941A>C

ENST00000371953.8:c.422A>C

ENST00000371953.7:c.422A>C

ENST00000498703.1:n.248A>C

ENST00000610634.1:c.320A>C

NM_000314.5:c.422A>C

NM_000314.6:c.422A>C

NM_001304717.2:c.941A>C

NM_001304718.1:c.-329A>C

NM_001304717.5:c.941A>C

NM_001304718.2:c.-329A>C

NM_000314.8:c.422A>C

Likely Pathogenic

Met criteria codes 3

PS3

PP2

PM2

Not Met criteria codes 23

BA1

BS1

BS4

BS3

BS2

BP5

BP7

BP4

BP3

BP1

BP2

PVS1

PS1

PS2

PS4

PP1

PP3

PP4

PM6

PM1

PM3

PM5

PM4

Evidence Links 0

Expert Panel

PTEN VCEP

Criteria Specification Information

Criteria Specifications for this VCEP

Evidence submitted by expert panel

PTEN VCEP

PTEN c.422A>C (p.His141Pro) 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 (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.

Met criteria codes

PS3			Matreyek: low abundance class (score = 0.133314163298692); Mighell: truncation-like range (score = -4.001333421)
PP2			This is a missense variant.
PM2			Absent in population databases.

Not Met criteria codes

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
PVS1			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			* Internal case data for Patient 1: female with adult-onset Lhermitte-Duclos disease, macrocephaly, multiple large fibroids at 40s (our variant identified through testing); mother with thyroid cancer, multiple large uterine fibroids at 40s (no genetic testing) * Internal case data for Patient 2: female with breast cancer at 30s, macrocephaly, polyposis, suspected neurofibroma(s); son with macrocephaly (64 cm), arteriovenous spinal epidural fistula at 20s (our variant identified through testing)
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			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

