

*Variant: NM\_000314.7(PTEN):c.610C>G (p.Pro204Ala)*

Version: 2.0

CA000535 [↗](#)

189415 (ClinVar) [↗](#)

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

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

**Inheritance Mode:** Autosomal dominant inheritance

**UID:** b5627c7e-eb93-4a97-95aa-58fd645e077c

**Approved on:** 2023-10-11

**Published on:** 2023-10-18

### *HGVS expressions*

#### **NM\_000314.7:c.610C>G**

NM\_000314.7(PTEN):c.610C>G (p.Pro204Ala)

NC\_000010.11:g.87952235C>G

CM000672.2:g.87952235C>G

NC\_000010.10:g.89711992C>G

CM000672.1:g.89711992C>G

NC\_000010.9:g.89701972C>G

NG\_007466.2:g.93797C>G

ENST00000700029.2:c.610C>G

ENST00000710265.1:c.610C>G

ENST00000472832.3:c.610C>G

ENST00000688158.2:n.1345C>G

ENST00000688922.2:c.\*440C>G

ENST00000700021.1:c.565C>G

ENST00000700022.1:c.493-5618C>G

ENST00000700023.1:n.1768C>G

ENST00000700024.1:n.2002C>G

ENST00000700025.1:n.1379C>G

ENST00000700029.1:c.444C>G

ENST00000706954.1:c.610C>G

ENST00000706955.1:c.\*645C>G

ENST00000686459.1:c.\*196C>G

ENST00000688158.1:c.\*721C>G

ENST00000688308.1:c.610C>G

ENST00000688922.1:c.531C>G

ENST00000693560.1:c.1129C>G

ENST00000371953.8:c.610C>G

ENST00000371953.7:c.610C>G

ENST00000472832.2:c.37C>G

NM\_000314.5:c.610C>G

NM\_000314.6:c.610C>G

NM\_001304717.2:c.1129C>G

NM\_001304718.1:c.19C>G

NM\_001304717.5:c.1129C>G

NM\_001304718.2:c.19C>G

NM\_000314.8:c.610C>G

**Pathogenic**

**Met criteria codes** 6

- PP2
- PP3
- PS3\_Moderate
- PM6\_Strong
- PM2\_Supporting
- PS4\_Supporting

**Not Met criteria codes** 20

- PS1
- PS2
- PP1
- PP4
- BA1
- PM1
- PM3
- PM5
- PM4
- PVS1
- BS2
- BS1
- BS4
- BS3
- BP5
- BP7
- BP4
- BP3
- BP1
- BP2

**Evidence Links** 2

**Expert Panel**

[PTEN VCEP](#)

**Criteria Specification Information**













- [Criteria Specification: ClinGen PTEN Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for PTEN Version 3.0.0](#)
- [Criteria Specification Approval History](#)
- [Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

**PTEN VCEP**



























**PTEN c.610C>G (p.Pro204Ala) meets criteria to be classified as pathogenic for PTEN Hamartoma Tumor syndrome in an autosomal dominant manner using modified ACMG criteria (ACMG Classification Rules Specified for PTEN Variant Curation version 3.0.0). Please see a summary of the rules and criteria codes in the "PTEN ACMG Specifications Summary" document (assertion method column). PM6\_S: One proband with presumed de novo occurrence (maternity/paternity not confirmed) for a patient with highly specific phenotype. (internal laboratory contributor SCV000222125.7) PS3\_M: Functional studies supportive of a damaging effect on the gene or gene product. Score of this variant = -1.675 ( $\leq -1.11$ ) on a high throughput phosphatase assay (PMID:29706350). PS4\_P: Proband(s) with phenotype specificity score of 1-1.5. (internal laboratory contributor SCV000222125.7) PM2\_P: 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. PP3: REVEL score > 0.7 (score of this variant = 0.881)**

**Met criteria codes**


<b>PP2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP3</b>			Score = 0.881
<b>PS3_Moderate</b>			Mighell score hypomorphic  Protein abundance score 0.52, "possibly low" range. <a href="#">PubMed:29785012</a> Score -1.675 in hypomorphic range. <a href="#">PubMed:29706350</a>
<b>PM6_Strong</b>			GDx #1 assumed de novo; patient with high phenotype specificity (meets PS4_P), data combined to upweight here.
<b>PM2_Supporting</b>			Absent gnomAD
<b>PS4_Supporting</b>			GDx #1: F child extreme macro and autism, peds score = 5, 1 phenotype point. GDx #2: M child macro NOS and DD; mother pos macro 59cm and thy nod CC score 10. Assumed de novo, PS4_P wrapped into PM6_S upweight. GDx #2:

male child with extreme macroceph (+5.5SD), delays. Inherited from HET mother with macrocephaly, thyroid nodules. Meets PS4\_P, 1 meiosis towards PP1.

### Not Met criteria codes

<b>PS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PS2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PP1</b>			1 meiosis from Gdx #2, described in PS4_P
<b>PP4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BA1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM5</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PM4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>PVS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BS3</b>			

No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

<b>BP5</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP7</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP4</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP3</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP1</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
<b>BP2</b>			No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline

#### Curation History

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