

Variant:
NM_000314.6(PTEN):c.-1195_-1184delAAGCCGCAGCAA

Version: 1.2

[CA163819](#)

[189433 \(ClinVar\)](#)

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

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

Inheritance Mode: Autosomal dominant inheritance

UUID: a4de2dba-9fd0-4e69-a9e5-2e6d4ec56af5

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HGVS expressions

NM_000314.6(PTEN):c.-1195_-1184delAAGCCGCAGCAA
NC_000010.11:g.87863274_87863285del
CM000672.2:g.87863274_87863285del
NC_000010.10:g.89623031_89623042del
CM000672.1:g.89623031_89623042del
NC_000010.9:g.89613011_89613022del
NG_007466.2:g.4837_4848del
NG_033079.1:g.5157_5168del
ENST00000706954.1:c.-17+632_-17+643del
ENST00000688308.1:c.-17+161_-17+172del
ENST00000445946.5:c.-794_-783del
ENST00000371953.7:c.-1196_-1185del
ENST00000445946.3:c.-794_-783del
NM_001126049.1:c.-794_-783del
NM_001126049.2:c.-794_-783del

Likely Benign

Met criteria codes **2**

BS4_Supporting **BP5**

Not Met criteria codes **23**

BS1 **BS3** **BS2** **BP7** **BP4** **BP3**
BP1 **BP2** **PM6** **PM2** **PS1**
PS2 **PS3** **PS4** **PM1** **PM3**
PM5 **PM4** **BA1** **PVS1** **PP1**
PP2 **PP3**

Evidence Links **6**

Expert Panel

[PTEN VCEP](#)

Criteria Specification Information **!**

[Criteria Specifications for this VCEP](#)

Evidence submitted by expert panel

PTEN VCEP

PTEN c.-1195del12 (NC_000010.10:g.89623031_89623042delAAGCCGCAGCAA) meets criteria to be classified as likely benign 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). BS4_P: Lack of segregation in affected members of one family. (Internal laboratory contributor SCV SCV000222146.7) BP5: Variant found in multiple cases with alternate molecular basis for disease. (Internal laboratory contributors SCV000222146.7, SCV000183826.5)

Met criteria codes

BS4_Supporting	✓	Consider BS4_P. Gdx internal data: identified in teenage F with macrocephaly. Also present in teenage sister with normal OFC, no PHTS cutaneous features. NEGATIVE in teenage sister with macrocephaly (OFC 62.2cm), overgrowth, early puberty, no PHTS-related skin findings. Mother, 40s, also positive for variant, no clinical info.
BP5	✓	Multiple co-occurrences from clinical laboratory contributors: SCV000222146.7, SCV000183826.5 - BRCA2, MSH6, MEN1, MLH1

Not Met criteria codes

BS1	✗	No code specific comments provided, please refer to the summary above or general recommendations provided in the guideline
BS3	✗	Per previous EP review, results from this study not applicable for variant interpretation/criteria application. PubMed:21532617 ↗
BS2	✗	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
PM2	✗	4/31,374 global gnomAD alleles, N insufficient in subpopulations for benign criteria.

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
PS3	✘	Per previous EP review, results from this study not applicable for variant interpretation/criteria application. PubMed:21532617
PS4	✘	Identified in a 19 year old Hispanic male jejunum carpeting of hamartomous polyps containing foveolar and gastric-type tissue with arborizing streaks of smooth muscle and minimal inflammation. NGS panel completed [APC, BMPR1A, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11, and TP53). Only finding was PTEN c.-1196_-1185del12. OFC 62cm but 6'2" and 225lbs, no dermatologic/other PHTS findings. CC score = 26, 0.5 phenotype specificity points. PubMed:28157521 Variant listed in Table S2 as a pathogenic mutation identified in 2 individuals. A 58.2-year-old and a 59.1-year-old. Participants met relaxed ICC operational criteria per subjects and methods section. No additional patient specific information reported. PubMed:21194675 Identified in a cohort of premenopausal breast cancer patients who underwent oophorectomy PubMed:28821472 Variant listed as identified in this study in Supplementary Table 4. No additional patient specific information reported. Patients met full or relaxed criteria for Cowden syndrome per methods section. PubMed:25669429 Identified in 2 individuals. The first, a 53 year old female, reported to have a history of breast at 51 and thyroid cancer at 42. The second, a 57 year old female, with a history of breast cancer at 52 and no history of thyroid cancer. PubMed:21532617 Identified in 2 females. A 35-year-old, non-hispanic white, non-Ashkenazi Jewish, no personal history of cancer, family history of gynecologic and prostate cancers (CC score = 0). A 57-year-old, non-hispanic white, non-Ashkenazi Jewish, personal history of endometrial cancer, family history of other cancers (CC Score = 1. PubMed:28495237
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
BA1	✘	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

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

Curation History [↗](#)

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