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**Genetic Testing for Cowden Syndrome -
PTEN Hamartoma Tumor Syndrome**

MP9488

Covered Service: Yes

**Prior Authorization
Required:** Yes

**Additional
Information:** Genetic testing is covered for a Prevea360 Health Plan member if the test results provide a direct medical benefit or guides reproductive decision-making for the Prevea360 Health Plan member. See [Genetic Testing MP9012](#) for additional information.

Pre and post-test genetic counseling is required for any individual undergoing genetic testing for hereditary breast and ovarian cancer syndromes.

Allowed once per lifetime in adults 18 years of age or older.

A first-degree relative is defined as an individual's parents, full siblings, and children.

A second-degree relative is defined as an individual's grandparents, grandchildren, aunts, uncles, nephews, nieces and half-siblings.

A third-degree relative is defined as first cousins, great-aunts, great-uncles, great-grandchildren, or great-grandparents.

Prevea360 Health Plan Medical Policy:

- 1.0 Genetic testing for **Cowden Syndrome (CS)** requires prior authorization through the Health Services Division. **PTEN** gene testing is considered medically necessary in individuals with a suspected or known clinical diagnosis of Cowden syndrome or Bannayan-Riley-Ruvalcaba syndrome (BRRS), or a known family history of a PTEN mutation who meet **ANY** of the following criteria:
 - 1.1 Individual from a family with a known PTEN mutation pathogenic/likely pathogenic variant
 - 1.2 Individual with a personal history of Bannayan-Riley-Ruvalcaba Syndrome
 - 1.3 Individual meeting clinical diagnostic criteria for Cowden Syndrome/PTEN Hamartoma Tumor Syndrome (PHTS)
 - 1.4 Individual **not** meeting clinical diagnostic criteria for CS/PHTS with a personal

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history of at least **ONE** of the following:

- 1.4.1 Adult Lhermitte-Duclos disease (cerebellar tumors)
 - 1.4.2 Autism spectrum disorder and macrocephaly
 - 1.4.3 Two (2) or more biopsy-proven trichilemmomas
 - 1.4.4 Two (2) or more major criteria (one (1) must be macrocephaly)
 - 1.4.5 Three (3) major criteria without macrocephaly
 - 1.4.6 One (1) major and three (3) or more minor criteria
 - 1.4.7 Four (4) or more minor criteria
- 1.5 At-risk individual with a relative with a clinical diagnosis of CS/PHTS or BRRS for whom testing has not been performed. The at-risk individual must have **EITHER** of the following:
- 1.5.1 Any one major criteria; **OR**
 - 1.5.2 Two minor criteria
- 1.6 PTEN pathogenic/likely variant detected by tumor profiling on any tumor type in the absence of germline analysis

***Criteria for PTEN genetic testing purposes are:**

Major	<p>Breast cancer</p> <p>Mucocutaneous lesions (ANY of the following):</p> <ul style="list-style-type: none"> ● One biopsy proven trichilemmoma ● Multiple palmoplantar keratoses ● Multifocal or extensive oral mucosal papillomatosis ● Multiple cutaneous facial papules (often verrucous) <p>Macrocephaly ($\geq 97^{\text{th}}$ percentile: 58 cm in adult females, 60 cm in adult males)</p> <p>Endometrial cancer</p> <p>Thyroid cancer (follicular)</p> <p>Multiple gastrointestinal hamartomas or ganglioneuromas</p> <p>Macular pigmentation of the glans penis</p>
Minor	<p>Colon Cancer</p> <p>Esophageal glycogenic acanthosis (≥ 3)</p>

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Minor	Thyroid structural lesions (e.g. adenoma, nodule(s), goiter) Testicular lipomatosis Vascular anomalies (including multiple intracranial developmental venous anomalies) Papillary or follicular variant of papillary thyroid cancer Intellectual disability (e.g. IQ ≤ 75) Autism spectrum disorder Single gastrointestinal hamartoma or ganglioneuroma Lipomas Renal cell carcinoma
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2.0 There is insufficient evidence to include fibrocystic disease of the breast, fibromas, and uterine fibroids as diagnostic criteria so these are considered experimental and investigational and therefore are not medically necessary.

CPT/HCPCS Codes Related to MP9488

* The list of codes (and their descriptors, if any) is provided for informational purposes only and may not be all inclusive or current. Listing of a code in this medical policy does not imply that the service described by the code is a covered or non-covered service. Benefit coverage for any service is determined by the member's policy of health coverage with Prevea360 Health Plan. Inclusion of a code above does not imply any right to reimbursement or guarantee claim payment. Other medical policies may also apply.

CPT Code	Description
0101U	Hereditary colon cancer disorders (eg, Lynch syndrome, PTEN hamartoma syndrome, Cowden syndrome, familial adenomatosis polyposis), genomic sequence analysis panel utilizing a combination of NGS, Sanger, MLPA, and array CGH, with mRNA analytics to resolve variants of unknown significance when indicated (15 genes [sequencing and deletion/duplication], EPCAM and GREM1 [deletion/duplication only])
0130U	Hereditary colon cancer disorders (eg, Lynch syndrome, PTEN hamartoma syndrome, Cowden syndrome, familial adenomatosis polyposis), targeted mRNA sequence analysis panel (APC, CDH1,

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	CHEK2, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, and TP53) (List separately in addition to code for primary procedure)
0235U	PTEN (phosphatase and tensin homolog) (eg, Cowden syndrome, PTEN hamartoma tumor syndrome), full gene analysis, including small sequence changes in exonic and intronic regions, deletions, duplications, mobile element insertions, and variants in non-uniquely mappable regions (e.g. Genomic Unity PTEN Analysis)
81321	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; full sequence analysis
81322	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; known familial variant
81323	PTEN (phosphatase and tensin homolog) (e.g., Cowden syndrome, PTEN hamartoma tumor syndrome) gene analysis; duplication/deletion variant
81403	Molecular Pathology Procedure Level 4
81435	Hereditary colon cancer disorders (e.g., Lynch syndrome, PTEN hamartoma syndrome, Cowden syndrome, familial adenomatosis polyposis); genomic sequence analysis panel, must include sequencing of at least 10 genes, including APC, BMPR1A, CDH1, MLH1, MSH2, MSH6, MUTYH, PTEN, SMAD4, and STK11
81436	Hereditary colon cancer disorders (e.g., Lynch syndrome, PTEN hamartoma syndrome, Cowden syndrome, familial adenomatosis polyposis); duplication/deletion analysis panel, must include analysis of at least 5 genes, including MLH1, MSH2, EPCAM, SMAD4, and STK11
81445	Targeted genomic sequence analysis panel, solid organ neoplasm, DNA analysis, and RNA analysis when performed, 5-50 genes (e.g., ALK, BRAF, CDKN2A, EGFR, ERBB2, KIT, KRAS, NRAS, MET, PDGFRA, PDGFRB, PGR, PIK3CA, PTEN, RET), interrogation for sequence variants and copy number variants or rearrangements, if performed

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	Committee/Source	Date(s)
Document Created:	Medical Policy Committee/Quality and Care Management Division	April 3, 2017
Revised:	Medical Policy Committee/Quality and Care Management Division	June 20, 2018
	Medical Policy Committee/Health Services Division	March 20, 2019
	Medical Policy Committee/Health Services Division	June 19, 2019
	Medical Policy Committee/Health Services Division	November 20, 2019
	Medical Policy Committee/Health Services Division	January 15, 2020
	Medical Policy Committee/Health Services Division	June 17, 2020
	Medical Policy Committee/Health Services Division	March 17, 2021
	Medical Policy Committee/Health Services Division	February 16, 2022
Reviewed:	Medical Policy Committee/Quality and Care Management Division	June 20, 2018
	Medical Policy Committee/Health Services Division	March 20, 2019
	Medical Policy Committee/Health Services Division	June 19, 2019
	Medical Policy Committee/Health Services Division	November 20, 2019
	Medical Policy Committee/Health Services Division	January 15, 2020
	Medical Policy Committee/Health Services Division	June 17, 2020
	Medical Policy Committee/Health Services Division	March 17, 2021
	Medical Policy Committee/Health Services Division	February 16, 2022

Published/Effective: 02/16/2022