Overview

Useful For
Confirming a diagnosis of PTEN hamartoma tumor syndrome, which includes Cowden syndrome, Bannayan-Riley-Ruvalcaba syndrome, Proteus syndrome, or Proteus-like syndrome

Identifying mutations in the PTEN gene

Additional Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>COLAB</td>
<td>Hereditary Colon Cancer CGH Array</td>
<td>Yes, (order FMTT)</td>
<td>Yes</td>
</tr>
</tbody>
</table>

Testing Algorithm
When this test is ordered, comparative genomic hybridization array will always be performed at an additional charge.

See Colonic Polyposis Syndromes Testing Algorithm in Special Instructions.

Special Instructions
- Molecular Genetics: Inherited Cancer Syndromes Patient Information
- Informed Consent for Genetic Testing
- Colonic Polyposis Syndromes Testing Algorithm
- Informed Consent for Genetic Testing (Spanish)

Method Name
Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing, Array (aCGH)

NY State Available
Yes

Specimen

Specimen Type
Varies

Shipping Instructions
Specimen preferred to arrive within 96 hours of draw.

Specimen Required
Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Specimen Type: Whole blood

Container/Tube:
Test Definition: PTENZ
PTEN Gene, Full Gene Analysis

Preferred: Lavender top (EDTA) or yellow top (ACD)

Acceptable: Any anticoagulant

Specimen Volume: 3 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.

Forms
1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:
   - Informed Consent for Genetic Testing (T576)
   - Informed Consent for Genetic Testing-Spanish (T826)
2. Molecular Genetics: Inherited Cancer Syndromes Patient Information (T519) in Special Instructions
3. If not ordering electronically, complete, print, and send a Gastroenterology and Hepatology Client Test Request (T728) with the specimen.

Specimen Minimum Volume
1 mL

Reject Due To
All specimens will be evaluated by Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
<th>Special Container</th>
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<tbody>
<tr>
<td>Varies</td>
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<tr>
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<td>Frozen</td>
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<td></td>
<td>Refrigerated</td>
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Clinical and Interpretive

Clinical Information
Germline mutations in the PTEN gene are associated with a rare collection of clinical syndromes referred to as PTEN hamartoma tumor syndrome (PHTS). This includes Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome (BRRS), Proteus syndrome (PS) and Proteus-like syndrome (PLS). Although each of these syndromes has its own unique features, all 4 appear to be associated with multiple hamartomatous lesions, vascular lesions, and macrocephaly. Affected individuals have an increased risk of cancer, including cancers of the breast, endometrium, thyroid, colon, and kidney. PHTS is an autosomal dominant disorder and penetrance is believed to be quite high.
CS is a multiple hamartoma syndrome associated with trichilemmomas, mucocutaneous papillomatous papules, and macrocephaly. Affected individuals are at an increased risk for breast, thyroid, and endometrial carcinoma.

BRRS is characterized by macrocephaly, intestinal hamartomas, lipomatosis, hemangiomatosis, and pigmented macules on the glans penis.

PS is associated with congenital malformations, overgrowth, macrocephaly, hyperostosis, connective tissue nevi, and epidermal nevi.

PLS refers to individuals who have features of PS, but do not meet diagnostic criteria.

Reference Values
An interpretive report will be provided.

Interpretation
All detected alterations are evaluated according to American College of Medical Genetics (ACMG) recommendations.(1) Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

Cautions
A small percentage of individuals who have a diagnosis of PTEN hamartoma tumor syndrome (PHTS) may have a mutation that is not identified by this method (eg, promoter mutations, deep intronic alterations). The absence of a mutation, therefore, does not eliminate the possibility of the diagnosis of PHTS. For testing asymptomatic individuals it is important to first document the presence of a PTEN gene mutation in an affected family member.

In some cases, DNA alterations of undetermined significance may be identified.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.

Clinical Reference

**Performance**

**Method Description**

Bidirectional sequence analysis is performed to test for the presence of a mutation in the promoter, all coding regions, and intron/exon boundaries of the *PTEN* gene. (Unpublished Mayo method)


**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Performed weekly; Varies

**Analytic Time**

14 days

**Maximum Laboratory Time**

20 days

**Specimen Retention Time**

Whole Blood: 2 weeks (if available) Extracted DNA: 3 months

**Performing Laboratory Location**

Rochester

**Fees and Codes**

**Fees**

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

**Test Classification**

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

**CPT Code Information**

81321-PTEN (phosphatase and tensin homolog) (eg. Cowden syndrome, PTEN hamartoma tumor syndrome gene analysis; full gene analysis)

Additional Tests

Hereditary Colon Cancer CGH Array
Test Definition: PTENZ
PTEN Gene, Full Gene Analysis

81228-Cytogenomic constitutional (genome-wide) microarray analysis; interrogation of genomic regions for copy number variants (eg, Bacterial Artificial Chromosome [BAC] or oligo-based comparative genomic hybridization [CGH] microarray analysis)

LOINC® Information

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