

Notification Date: October 8, 2024 Effective Date: December 2, 2024

PTEN Hamartoma Tumor Syndrome, *PTEN* Full Gene Analysis, Varies

Test ID: PTNZ

Explanation: On the effective date, this assay will incorporate changes to the specimen requirements. Whole blood stability guidance will be provided, and *any anticoagulant* will no longer be accepted. Anticoagulants will be limited to those listed below. Saliva will also be added as an acceptable specimen type.

Current 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

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 whole blood specimen in original tube. **Do** not aliquot.

Specimen Stability Information: Ambient (preferred) 4 days/Refrigerated

New 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

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

Acceptable: Sodium heparin

Specimen Volume: 3 mL

Collection Instructions:

1. Invert several times to mix blood.

2. Send whole blood specimen in original tube. **Do** not aliquot.

Specimen Stability Information:

Ambient/Refrigerated/Frozen, 96 hours (preferred); extraction will be attempted for samples received after 96 hours and DNA yield will be evaluated to determine if testing may proceed

Additional Information: To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted.

Testing may be canceled if DNA requirements are inadequate. Specimen Type: Saliva Patient Preparation: Patient should not eat, drink, smoke, or chew gum 30 minutes prior to collection. Supplies: Saliva Collection Kit (T786) -ORAcollect OCD-100 Swab Specimen Volume: 1 Swab Collection Instructions: Collect and send specimen per kit instructions. Specimen Stability Information: Ambient (preferred), Refrigerated acceptable; 30 days Additional information: Due to lower quantity/quality of DNA yielded from saliva, some aspects of the test may not perform as well as DNA extracted from a whole blood sample. When applicable, specific gene regions that were unable to be interrogated will be noted in the report. Alternatively, additional specimen may be required to complete testing.