

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

Birt-Hogg-Dube Syndrome, *FLCN*, Full Gene Analysis, Varies

Test ID: BHDZ

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 New Specimen Required Patient Preparation: A previous bone marrow Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow testing patients who have received a bone marrow transplant. transplant. Specimen Type: Whole blood Specimen Type: Whole blood Preferred: Lavender top (EDTA) or yellow top Preferred: Lavender top (EDTA) or yellow top (ACD) (ACD) Acceptable: Any anticoagulant Acceptable: Sodium heparin Specimen Volume: 3 mL Specimen Volume: 3 mL **Collection Instructions: Collection Instructions:** 1. Invert several times to mix blood. 1. Invert several times to mix blood. 2. Send whole blood specimen in original tube. Do 2. Send whole blood specimen in original tube. **Do** not aliquot. not aliquot. Specimen Stability Information: Ambient **Specimen Stability Information:** (preferred) 4 days/Refrigerated 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.