

Fabry Disease, Full Gene Analysis, Varies

Test ID: FABRZ

Explanation: On the effective date, this assay will incorporate changes to the Specimen Required, as shown below. The Whole Blood specimen type will no longer accept *any anticoagulant* and will be limited to the anticoagulants shown below. Additionally, stability guidance for Whole Blood will be provided.

| Current Specimen Required | New Specimen Required |
|--|--|
| <p>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.</p> <p>Submit only 1 of the following specimens:</p> <p>Preferred: Specimen Type: Whole blood Container/Tube: Preferred: Lavender top (EDTA) tube or yellow top (ACD) tube Acceptable: Any anticoagulant Specimen Volume: 3 mL Collection Instructions: 1. Invert several times to mix blood. 2. Send specimen in original tube. Specimen Stability Information: Ambient (preferred)/Refrigerated</p> <p>Acceptable: Specimen Type: Blood spot Supplies: Card - Blood Spot Collection (Filter Paper) (T493) Container/Tube: Preferred: Collection card (Whatman Protein Saver 903 Paper) Acceptable: Ahlstrom 226 filter paper, or Blood Spot Collection Card Specimen Volume: 2 to 5 Blood spots on collection card Collection Instructions: 1. An alternative blood collection option for a patient older than 1 year of age is finger stick. 2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.</p> | <p>Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.</p> <p>Submit only 1 of the following specimens:</p> <p>Preferred: Specimen Type: Whole blood Container/Tube: Preferred: Lavender top (EDTA) tube or yellow top (ACD) tube Acceptable: 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 4 days/Refrigerated 4 days/Frozen 4 days Additional Information: 1. Specimens are preferred to be received within 4 days of collection. Extraction will be attempted for specimens received after 4 days, and DNA yield will be evaluated to determine if testing may proceed. 2. 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</p> <p>Acceptable: Specimen Type: Blood spot Supplies: Card - Blood Spot Collection (Filter Paper) (T493) Container/Tube:</p> |

3. Do not expose specimen to heat or direct sunlight.

4. Do not stack wet specimens.

5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. For collection instructions, see [Blood Spot Collection Instructions](#) in Special Instructions.

2. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777) in Special Instructions.

3. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800) in Special Instructions.

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper, or Blood Spot Collection Card

Specimen Volume: 2 to 5 Blood spots on collection card

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year of age is finger stick.

2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.

3. Do not expose specimen to heat or direct sunlight.

4. Do not stack wet specimens.

5. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Additional Information:

1. Due to lower concentrations of DNA yielded from blood spots, 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 needed to complete testing.

2. For collection instructions, see [Blood Spot Collection Instructions](#)

3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)

4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

Questions

Contact Michelle Raths, Laboratory Resource Coordinator at 800-533-1710.