

Hereditary Erythrocytosis Gene Panel, Next-Generation Sequencing, Varies

Test ID: NHEP

Explanation: On the effective date, NHEP will begin accepting Skin Biopsies and Cultured Fibroblasts, in addition to the already acceptable Whole Blood specimen. The Testing Algorithm and Reflex Testing will be updated, as shown below.

Current Testing Algorithm
<p>This evaluation is recommended for patients presenting with lifelong elevation in hemoglobin or hematocrit, usually with a positive family history of similar symptoms. Polycythemia vera should be excluded prior to testing as it is much more common than hereditary erythrocytosis and can be present even in young patients. A JAK2 V617F or JAK2 exon 12 variant should not be present. More sensitive, variant-specific testing for JAK2 V617F is highly recommended prior to ordering this test. Additionally, alpha and beta chain high-oxygen affinity hemoglobin variants should be excluded prior to ordering this test panel.</p>

New Testing Algorithm
<p>This evaluation is recommended for patients presenting with lifelong elevation in hemoglobin or hematocrit, usually with a positive family history of similar symptoms. Polycythemia vera should be excluded prior to testing as it is much more common than hereditary erythrocytosis and can be present even in young patients. A JAK2 V617F or JAK2 exon 12 variant should not be present. More sensitive, variant-specific testing for JAK2 V617F is highly recommended prior to ordering this test. Additionally, alpha and beta chain high-oxygen affinity hemoglobin variants should be excluded prior to ordering this test panel.</p> <p>If skin biopsy (fresh) is received, fibroblast culture will be added at an additional charge. If viable cells are not obtained, the client will be notified.</p>

Current Reflex Testing
None

New Reflex Testing			
Test ID	Reporting Name	Available Separately	Always Performed
CULFB	Fibroblast Culture for Genetic Test	Yes	No

Current Specimen Required
<p>Specimen Type: Whole blood</p> <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>

New Specimen Required
<p>Submit only 1 of the following specimens:</p> <p>Specimen Type: Whole blood</p> <p>Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with whole blood testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.</p>

Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD)
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

Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD)
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

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing, Chorionic Villi/Products of Conception/Tissue. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Cultured fibroblast

Container/Tube: T-25 flask

Specimen Volume: 2 Flasks

Collection Instructions: Submit confluent cultured fibroblast cells from a skin biopsy from another laboratory. Cultured cells from a prenatal specimen will not be accepted.

Specimen Stability Information: Ambient (preferred)/Refrigerated (<24 hours)

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing, Chorionic Villi/Products of Conception/Tissue. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Questions

Contact Connie Penz, Laboratory Resource Coordinator at 800-533-1710.