
Hereditary Erythrocytosis Mutations, Whole Blood

Test ID: HEMP

Explanation:

This test will become obsolete on March 24, 2026. The recommended alternative is listed below.

Recommended Alternative Test:

Hereditary Erythrocytosis Gene Panel, Next-Generation Sequencing, Varies

Test ID: NHEP

Genetics Information:

This test utilizes next-generation sequencing to detect single nucleotide and copy number variants in 24 genes associated with hereditary erythrocytosis and polycythemia: *ACO1*, *ANKRD26*, *BHLHE41*, *BPGM*, *CYB5A*, *CYB5R3*, *EGLN1*, *EGLN2*, *EGLN3*, *EPAS1*, *EPO*, *EPOR*, *GFI1B*, *HIF1A*, *HIF1AN*, *HIF3A*, *JAK2*, *KDM6A*, *PFKM*, *PIEZO1*, *PKLR*, *SH2B3*, *SOCS3*, and *VHL*. See Method Description for additional details.

Identification of a disease-causing variant may assist with diagnosis, prognosis, clinical management, recurrence risk assessment, familial screening, and genetic counseling for hereditary erythrocytosis.

Reflex Tests:

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

Methodology:

Sequence Capture and Targeted Next-Generation Sequencing (NGS) followed by Polymerase Chain Reaction (PCR) and Sanger Sequencing

Reference Values:

An interpretive report will be provided.

Specimen Requirements:

Submit only 1 of the following specimens:

Specimen Type: Whole blood

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with whole blood testing. For information about testing patients who have received a bone marrow transplant, call 800-533-1710.

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 4 days**Additional Information:** To ensure minimum volume and concentration of DNA are met, the requested volume must be submitted. Testing may be canceled if DNA requirements are inadequate.**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) < 24 hours/Ambient < 24 hours**Additional Information:**

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are 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) < 24 hours/Refrigerated <24 hours**Additional Information:**

1. Specimens are preferred to be received within 24 hours of collection. Culture and extraction will be attempted for specimens received after 24 hours and will be evaluated to determine if testing may proceed.
2. A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks are required to culture fibroblasts before genetic testing can occur.

Specimen Stability Information:

Specimen Type	Temperature	Varies
Varies	Varies	

CPT Code:

81404

81405

81479

81479 (if appropriate for government payers)

Day(s) Performed: Varies **Report Available:** 28 to 42 days**Questions**

Contact Melissa Lonzo, Laboratory Resource Coordinator, at 800-533-1710.