
Reporting Title: Erythrocytosis Focused Panel, NGS**Performing Location:** Rochester**Ordering Guidance:**

Polycythemia vera and acquired causes of erythrocytosis should be excluded before ordering this evaluation.

For a complete evaluation including hemoglobin electrophoresis testing and hereditary erythrocytosis variant analysis of the most common gene regions associated with hereditary erythrocytosis in an algorithmic fashion, order REVE2 / Erythrocytosis Evaluation, Blood. See [Erythrocytosis Genotyping Comparison Chart](#) for a comparison of erythrocytosis testing options.

The hemoglobin genes, *HBA1/HBA2* and *HBB*, are not interrogated in this assay.

Multiple gene panels are available. For more information see [NHEP and Subpanel Comparison Gene List](#).

Customization of this panel and single gene analysis for any gene present on this panel are available. For more information see CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies.

Targeted testing for familial variants (also called site-specific or known variants testing) is available for the genes on this panel. See FMTT / Familial Variant, Targeted Testing, Varies. To obtain more information about this testing option, call 800-533-1710.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Necessary Information:

- [Erythrocytosis Patient Information](#) is required. Testing may proceed without the patient information, however, the information aids in providing a more thorough interpretation. Ordering providers are strongly encouraged to fill out the form and send with the specimen.
- If form not provided, include the following information with the test request: clinical diagnosis, pertinent clinical history (ie, complete blood cell count results and relevant clinical notes), and differentials based on clinical presentation and/or laboratory findings.

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. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 3 mL

Collection Instructions:

- 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. 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. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Forms:

- 1. [Erythrocytosis Patient Information \(T694\)](#) is required.
- 2. **New York Clients-Informed consent is required.** Document on the request form or electronic order that a copy is on file. The following documents are available:
 - [Informed Consent for Genetic Testing \(T576\)](#)
 - [Informed Consent for Genetic Testing \(Spanish\) \(T826\)](#)
- 3. [If not ordering electronically, complete, print, and send a Benign Hematology Test Request \(T755\)](#) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
618992	Test Description	Alphanumeric		62364-5
618993	Specimen	Alphanumeric		31208-2
618994	Source	Alphanumeric		31208-2
618995	Result Summary	Alphanumeric		50397-9
618996	Result	Alphanumeric		82939-0
618997	Interpretation	Alphanumeric		59465-5
618998	Additional Results	Alphanumeric		82939-0
618999	Resources	Alphanumeric		99622-3

619000	Additional Information	Alphanumeric		48767-8
619001	Method	Alphanumeric		85069-3
619002	Genes Analyzed	Alphanumeric		82939-0
619003	Disclaimer	Alphanumeric		62364-5
619004	Released By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

Supplemental

CPT Code Information:

- 81404
- 81479
- 81479 (if appropriate for government payers)

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
CULFB	Fibroblast Culture for Genetic Test	1	88233	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.