MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: October 30, 2012
EFFECTIVE DATE: November 26, 2012

Hereditary Erythrocytosis Mutations
Test ID: HEMP

USEFUL FOR: The definitive evaluation of an individual with JAK2 negative erythrocytosis associated with lifelong sustained increased RBC mass, elevated RBC count, hemoglobin or hematocrit.

PROFILE INFORMATION:

Test ID	Reporting Name	Available Separately	Always Performed
MINT	Molecular Interpretation	No	Yes
EPOR	EPOR Gene, Mutation Analysis, B	No	Yes
HIF2A	HIF2A Gene, Mutation Analysis, B	No	Yes
PHD2	PHD2 Gene, Mutation Analysis, B	No	Yes

REFLEX TESTS:

Test ID	Reporting Name	Available Separately	Always Performed
VHLSP	VHL Full Gene Analysis	Yes	No

METHODOLOGY: Polymerase Chain Reaction (PCR) Amplification/Sanger Sequence Analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS:
Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions: Send specimen in original tube.
Forms:
1. Patient Information Sheet-Erythrocytosis Patient Information Sheet in Special Instructions
2. **New York Clients-Informed consent is required.** Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Whole Blood	Refrigerated	30 days

CAUTIONS:
- Polycythemia vera and acquired causes of erythrocytosis should be excluded before ordering this evaluation. The p50 value should be normal.
- This test is not intended for prenatal diagnosis.
- Certain mutations have no clinical manifestations and, in essence, are clinically benign. Correlation with all relevant clinical information is necessary to provide appropriate patient care.

FEE: Please contact your Regional Manager for your account's fee information.

CPT CODES:
Molecular Interpretation
83890-Automated extraction
83912-Interpretation and Report

EPOR Gene, Mutation Analysis, B
83900-Amplification, target, multiplex, first 2 nucleic acid sequences
83904 x 4-Mutation identification by DNA sequencing, single segment, each segment
83909 x 4-Separation and identification by high resolution technique

HIF2A Gene, Mutation Analysis, B
83900-Amplification, target, multiplex, first 2 nucleic acid sequences
83904 x 4-Mutation identification by DNA sequencing, single segment, each segment
83909 x 4-Separation and identification by high resolution technique

PHD2 Gene, Mutation Analysis, B
83900-Amplification, target, multiplex, first 2 nucleic acid sequences
83901 x 3-Amplification, target, multiplex, each additional nucleic acid sequence beyond 2
83904 x 12-Mutation identification by DNA sequencing, single segment, each segment
83909 x 12-Separation and identification by high resolution technique

VHL Full Gene Analysis (if appropriate)
81404-VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), full gene sequence
81403-VHL (von Hippel-Lindau tumor suppressor) (eg, von Hippel-Lindau familial cancer syndrome), deletion/duplication analysis (if appropriate)

DAY(S) SET UP: Monday through Friday **ANALYTIC TIME**: 10 days

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Julie Breider, MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710