

Reporting Title: Hereditary Erythrocytosis Mut, B
Performing Location: Rochester

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 Type	Temperature	Time
Whole blood	Refrigerated	30 days

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
34648	Molecular Interpretation	Alphanumeric		In Process
35000	Reviewed By	Numeric		In Process
34645	EPOR Gene Sequencing Result	Alphanumeric		In Process
34647	HIF2A Gene Sequencing Result	Alphanumeric		In Process
34646	PHD2 Gene Sequencing Result	Alphanumeric		In Process

Components:

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

CPT Code Information:

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)
 For nonparticipating payers:
 Von Hippel-Lindau (VHL) Gene, Full Gene Analysis
 83892 x 2-Enzymatic digestion
 83894-Separation by gel electrophoresis
 83900-Amplification, target, multiplex, first 2 nucleic acid sequences
 83901 x 4-Amplification, target, multiplex, each additional nucleic acid sequence beyond 2
 83912-Interpretation and report
 VHL Gene Sequencing
 83909 x 6
 VHL Deletion Detection
 83900-Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)
 83901 x 14-Amplification, target, multiplex, each additional nucleic acid sequence beyond 2 (if appropriate)
 83909-Separation and identification by high-resolution technique (if appropriate)
 83914-Mutation identification by enzymatic ligation or primer extension, single segment, each segment (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
VHLS	VHL Full Gene Analysis		Profile	No	Yes
ENDEX	DNA Extraction	1	83891	Yes	No
VHDD	VHL Deletion Detection		Profile	No	Yes (order VHLD)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
VHLSP	28626	Reason for Referral	Alphanumeric		42349-1
VHLSP	28627	Method	Alphanumeric		In Process
VHLSP	28628	Result	Alphanumeric		N/A
VHLSP	28629	Interpretation	Alphanumeric		69047-9
VHLSP	28630	Comment	Alphanumeric		In Process
VHLSP	28631	Reviewed By	Numeric		N/A
VHLSP	VHLSQ	Sequencing	Alphanumeric		In Process
ENDEX	ENDEX	DNA Extraction	Numeric		N/A
VHDD	VHDD	VHL Deletion Detection	Numeric		In Process

Reference Values:

An interpretive report will be provided.