

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

Ordering Guidance:
For a complete evaluation including hemoglobin electrophoresis testing and hereditary erythrocytosis variant analysis in an algorithmic fashion, order REVE2 / Erythrocytosis Evaluation, Blood.

This test does not provide a serum erythropoietin (EPO) level. If EPO testing is desired, order EPO / Erythropoietin, Serum.

Necessary Information:
[Erythrocytosis Patient Information](#) (T694) is strongly recommended, but not required, to be filled out and sent with the specimen. This information aids in providing a more thorough interpretation of test results. Ordering providers are strongly encouraged to complete the form and send it with the specimen.

Specimen Requirements:
Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot.**

Forms:
1. **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 in Special Instructions:
-[Informed Consent for Genetic Testing](#) (T576)
-[Informed Consent for Genetic Testing-Spanish](#) (T826)
2. [Erythrocytosis Patient Information](#) (T694)
3. If not ordering electronically, complete, print, and send a [Benign Hematology Test Request Form](#) (T755) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	30 days	
	Ambient	14 days	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
34645	EPOR Gene Sequencing Result	Alphanumeric		82939-0
34646	PHD2 Gene Sequencing Result	Alphanumeric		82939-0
34647	HIF2A Gene Sequencing Result	Alphanumeric		82939-0
34648	Molecular Interpretation	Alphanumeric		69047-9
35000	Reviewed By	Alphanumeric		18771-6

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

Supplemental Report:

No

Components:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
MINT	Molecular Interpretation			Yes	No
EPOR	EPOR Gene, Mutation Analysis, B	1	81479	Yes	No
HIF2A	HIF2A Gene, Mutation Analysis, B	1	81479	Yes	No
PHD2	PHD2 Gene, Mutation Analysis, B	1	81479	Yes	No

CPT Code Information:

81479

Reflex Tests:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
BPGMM	BPGM Full Gene Sequencing	1	81479	Yes	Yes
VHLE	VHL Gene Erythrocytosis Mutations	1	81404	Yes	No, (Order VHLZZ)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
VHLE	37886	Known Mut Reason for Referral	Alphanumeric		42349-1
VHLE	37840	Result Summary	Alphanumeric		50397-9
VHLE	37841	Result	Alphanumeric		82939-0
VHLE	37842	Interpretation	Alphanumeric		69047-9
VHLE	37843	Additional Information	Alphanumeric		48767-8
VHLE	37844	Specimen	Alphanumeric		31208-2
VHLE	37845	Source	Alphanumeric		31208-2
VHLE	37846	Released By	Alphanumeric		18771-6
BPGMM	37111	BPGM Gene Sequencing Result	Alphanumeric		No LOINC Needed
BPGMM	37112	BPGM Interpretation	Alphanumeric		69047-9

Reference Values:

An interpretive report will be provided.