

## Erythrocytosis Evaluation, Blood

**Test ID:** REVE2

**Explanation:** On the effective date, the following changes will be made to the Reflex Tests. The Testing Algorithm will be updated to reflect the changes.

Testing to be removed from Reflex Tests:

HEMP / Hereditary Erythrocytosis Mutations, Whole Blood

BPGMM / 2,3-Bisphosphoglycerate Mutase, Full Gene Sequencing Analysis, Varies

VHLE / *VHL* Gene, Erythrocytosis, Mutation Analysis, Varies

Testing to be added to Reflex Tests:

NHEP / Hereditary Erythrocytosis Gene Panel, Next-Generation Sequencing, Varies

### Current Algorithm

This is a consultative evaluation in which the case will be evaluated at Mayo Clinic Laboratories, the appropriate tests will be performed at an additional charge, and the results interpreted.

This profile evaluates for hereditary (congenital) causes of erythrocytosis. Symptoms should be long-standing or familial in nature. All cases will be tested for hemoglobin variants (cation exchange high performance liquid chromatography, capillary electrophoresis, and mass spectrometry) with an interpretative report. Additional testing is guided in a reflexive manner and may include molecular testing of the *HBA1/HBA2*, *HBB*, *EPOR*, *VHL*, *EGLN1(PHD2)*, *EPAS1(HIF2a)*, and *BPGM* genes, among others, as appropriate. For more information see [Erythrocytosis Evaluation Testing Algorithm](#).

If any of the following molecular tests are performed, an additional consultative interpretation that summarizes all testing will be provided to incorporate subsequent results into an overall evaluation:

-WAGDR / Alpha Globin Cluster Locus Deletion/Duplication, Blood

-WASQR / Alpha -Globin Gene Sequencing, Blood

-WBSQR / Beta-Globin Gene Sequencing, Blood

-WBGDR / Beta-Globin Gene Cluster Deletion/Duplication, Blood

-WGSQR / Gamma-Globin Full Gene Sequencing, Varies

Additional reflex tests are performed if the hemoglobin testing does not explain the patient's phenotype/hereditary erythrocytosis. Each of the following reflex tests contains an individual interpretative report.

-BPGMM / 2,3-Bisphosphoglycerate Mutase, Full Gene Sequencing Analysis, Varies

-HEMP / Hereditary Erythrocytosis Mutations, Whole Blood

-VHLE / *VHL* Gene, Erythrocytosis, Mutation Analysis, Varies

## New Algorithm

This is a consultative evaluation in which the case will be evaluated at Mayo Clinic Laboratories, the appropriate tests will be performed at an additional charge, and the results interpreted.

This profile evaluates for hereditary (congenital) causes of erythrocytosis. Symptoms should be long-standing or familial in nature. All cases will be tested for hemoglobin variants (cation exchange high performance liquid chromatography, capillary electrophoresis, and mass spectrometry) with an interpretative report. Additional testing is guided in a reflexive manner and may include molecular testing of the *HBA1/HBA2* and *HBB* genes, among others, as appropriate. For more information see [Erythrocytosis Evaluation Testing Algorithm](#).

If the hemoglobin testing results do not explain the patient's phenotype/hereditary erythrocytosis, the next-generation sequencing gene panel for hereditary erythrocytosis (NHEP) will be performed as a reflex at an additional charge. An individual interpretive report will be provided.

If any of the following molecular tests are performed, an additional consultative interpretation that summarizes all testing will be provided to incorporate subsequent results into an overall evaluation:

- WAGDR / Alpha Globin Cluster Locus Deletion/Duplication, Blood
- WASQR / Alpha -Globin Gene Sequencing, Blood
- WBSQR / Beta-Globin Gene Sequencing, Blood
- WBGDR / Beta-Globin Gene Cluster Deletion/Duplication, Blood
- WGSQR / Gamma-Globin Full Gene Sequencing, Varies

Current Reflex Tests	
Test ID	Reporting Name
SDEX	Sickle Solubility, B
<b>HEMP</b>	<b>Hereditary Erythrocytosis Mut, B</b>
IEF	Isoelectric Focusing, B
UNHB	Hb Stability, B
HPFH	Hb F Distribution, B
WASQR	Alpha Globin Gene Sequencing, B
WBSQR	Beta Globin Gene Sequencing, B
WGSQR	Gamma Globin Full Gene Sequencing
<b>BPGMM</b>	<b>BPGM Full Gene Sequencing</b>
REVE0	Erythrocytosis Summary Interp
WAGDR	Alpha Globin Clustr Locus Del/Dup,B
WBGDR	Beta Globin Gene Cluster, Del/Dup,B
<b>VHLE</b>	<b>VHL Gene Erythrocytosis Mutations</b>

New Reflex Tests	
Test ID	Reporting Name
SDEX	Sickle Solubility, B
IEF	Isoelectric Focusing, B
UNHB	Hb Stability, B
HPFH	Hb F Distribution, B
WASQR	Alpha Globin Gene Sequencing, B
WBSQR	Beta Globin Gene Sequencing, B
WGSQR	Gamma Globin Full Gene Sequencing
REVE0	Erythrocytosis Summary Interp
WAGDR	Alpha Globin Clustr Locus Del/Dup,B
WBGDR	Beta Globin Gene Cluster, Del/Dup,B
<b>NHEP</b>	<b>Erythrocytosis Full Panel, NGS</b>

## Questions

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