

Overview

Useful For

Incorporating and summarizing subsequent results into an overall evaluation if 1 or more molecular tests are reflexed on the REVE2 / Erythrocytosis Evaluation, Blood

Testing Algorithm

This test is an additional consultative interpretation that summarizes all testing, as well as any pertinent clinical information, and will be provided after test completion to incorporate subsequent molecular results into an overall evaluation if 1 or more of the following molecular tests are reflexed on the REVE2 / Erythrocytosis Evaluation, Blood:

- ATHAL / Alpha-Globin Gene Analysis, Varies
- WASQR / Alpha Globin Gene Sequencing, Blood
- WBSQR / Beta-Globin Gene Sequencing, Blood
- WBDDR / Beta-Globin Cluster Locus Deletion/Duplication, Blood
- WGSQR / Gamma-Globin Full Gene Sequencing, Varies
- HEMP / Hereditary Erythrocytosis Mutations, Whole Blood
- BPGMM / 2,3-Bisphosphoglycerate Mutase, Full Gene Sequencing Analysis, Varies

This summary interpretation is in addition to interpretations of individual components, including the erythrocytosis evaluation, hereditary erythrocytosis mutations, and von Hippel-Lindau tests.

Method Name

Only orderable as a reflex. For more information see REVE2 / Erythrocytosis Evaluation, Blood.

Medical Interpretation

NY State Available

Yes

Specimen

Specimen Type

Whole Blood EDTA

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood EDTA	Refrigerated		

Clinical & Interpretive

Clinical Information

The etiology of congenital (inherited) erythrocytosis can be due to one of several abnormalities. This includes high oxygen affinity hemoglobin variants, genetic variants in the erythropoietin receptor gene, genetic variants in the genes involved in the oxygen-sensing pathway (*PHD2/EGLN1*, *HIF2A/EPAS1*, *VHL*) or *BPGM* variants causing 2,3-BPG (2,3-bisphosphoglycerate) deficiency. To determine the underlying abnormality frequently requires molecular testing. A summary interpretation that incorporates all of the testing performed is beneficial to the ordering clinician.

Reference Values

Only orderable as a reflex. For more information see REVE2 / Erythrocytosis Evaluation, Blood.

An interpretive report will be provided.

Interpretation

An interpretive report will be provided.

Cautions

No significant cautionary statements

Clinical Reference

1. Patnaik MM, Tefferi A. The complete evaluation of erythrocytosis: congenital and acquired. *Leukemia*. 2009;23(5):834-844
2. Percy MJ, Lee FS. Familial erythrocytosis: molecular links to red blood cell control. *Haematologica*. 2008;93(7):963-967
3. Maran J, Prchal J: Polycythemia and oxygen sensing. *Pathol Biol (Paris)*. 2004 Jun;52(5):280-284
4. Merchant SH, Oliveira JL, Hoyer JD, Viswanatha DS: Erythrocytosis. In: His ED, ed. *Hematopathology*. 2nd ed. Elsevier Saunders; 2012:722-723
5. Hartevelde CL, Higgs DR. Alpha-thalassemia. *Orphanet J Rare Dis*. 2010;5:13
6. Thein SL. The molecular basis of beta-thalassemia. *Cold Spring Harb Perspect Med*. 2013 1;3(5):a011700
7. Crowley MA, Mollan TL, Abdulmalik OY, et al. A hemoglobin variant associated with neonatal cyanosis and anemia. *N Engl J Med*. 2011;364(19):1837-1843
8. Kipp BR, Roellinger SE, Lundquist PA, Highsmith WE, Dawson DB. Development and clinical implementation of a combination deletion PCR and multiplex ligation-dependent probe amplification assay for detecting deletions involving the human alpha-globin gene cluster. *J Mol Diagn*. 2011;13(5):549-557. doi:10.1016/j.jmoldx.2011.04.001
9. Hein MS, Oliveira JL, Swanson KC, et al: Large deletions involving the beta globin gene complex: genotype-phenotype correlation of 119 cases. *Blood*. 2015;126:3374

Performance**Method Description**

A hematopathologist evaluates all of the testing performed and a summary interpretive report is added.

PDF Report

No

Day(s) Performed

Monday through Friday

Report Available

3 to 25 days

Specimen Retention Time

28 days

Performing Laboratory Location

Rochester

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

Test Classification

Not Applicable

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
REVE0	Erythrocytosis Summary Interp	59465-5

Result ID	Test Result Name	Result LOINC® Value
608094	Erythrocytosis Summary Interp	59465-5
608116	Reviewed By	18771-6