

Test Definition: REVE0

Erythrocytosis Summary Interpretation

Overview

Useful For

Incorporating and summarizing subsequent results into an overall interpretation for the REVE2 / Erythrocytosis Evaluation, Blood

Testing Algorithm

When 1 or more molecular tests are added to the REVE2 / Erythrocytosis Evaluation, Blood, then this test is also added as consultative interpretation that summarizes the testing performed as well as any pertinent clinical information. This summary is in addition to interpretations that may be provided for each component. This will be provided after additional testing is complete in order to incorporate subsequent results into an overall evaluation.

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 testing performed is beneficial to the ordering clinician.

Reference Values



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An interpretive report will be provided.

Interpretation

An interpretive report will be provided that summarizes all testing as well as any pertinent clinical information.

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
- Percy MJ, Lee FS. Familial erythrocytosis: molecular links to red blood cell control. Haematologica. 2008;93(7):963-967
 Maran J, Prchal J. Polycythemia and oxygen sensing. Pathol Biol (Paris). 2004;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. Harteveld CL, Higgs DR. Alpha-thalassemia. Orphanet J Rare Dis. 2010;5:13

6. Thein SL. The molecular basis of beta-thalassemia. Cold Spring Harb Persepct 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 results from the testing performed, and a summary interpretation is provided.

PDF Report No

Day(s) Performed Monday through Friday

Report Available 3 to 25 days

Specimen Retention Time 28 days

Performing Laboratory Location



Erythrocytosis Summary Interpretation

Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees

- Authorized users can sign in to <u>Test Prices</u> 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
REVEO	Erythrocytosis Summary Interp	59465-5

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