

Overview

Useful For

Incorporating and summarizing subsequent molecular results into an overall evaluation if 1 or more molecular tests are reflexed on the THEV1 / Thalassemia and Hemoglobinopathy 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 THEV1 / Thalassemia and Hemoglobinopathy 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

This summary is in addition to interpretation of individual components of the Thalassemia Evaluation, such as the hemoglobin electrophoresis and molecular tests.

Method Name

Only orderable as a reflex. For more information see THEV1 / Thalassemia and Hemoglobinopathy 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 and Interpretive

Clinical Information

Some hemoglobin disorders can be very complex and involve abnormalities of the alpha, beta, delta, and gamma genes. These abnormalities can be due to, not only to point variants, but also deletions within 1 or more globin genes.

Multiple genetic variants can be seen in the same patient, and molecular testing is necessary to fully evaluate such cases.

A summary interpretation that incorporates all of the testing performed is beneficial to the ordering physician.

Reference Values

Only orderable as a reflex. For more information see THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Blood.

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. OMIM: 141800 Hemoglobin-alpha locus 1; HBA1. Updated 11/01/2019. Accessed 10/19/2020. Available at www.omim.org/entry/141800?search=141800&highlight=141800
2. OMIM: 141900 Hemoglobin-beta locus; HBB. Updated 11/14/2019. Accessed 10/19/2020. Available at www.omim.org/entry/141900?search=141900&highlight=141900
3. Kipp BR, Roellinger SE, Lundquist PA, Highsmith WE, Dawson DB et al: 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 Sep;13(5):549-557 doi: 10.1016/j.jmoldx.2011.04.001
4. Thom CS, Dickson CF, Gell DA, Weiss MJ: Hemoglobin variants: biochemical properties and clinical correlates. *Cold Spring Harb Perspect Med.* 2013;3(3):a011858
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. 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

14 days

Performing Laboratory Location

Rochester

Fees and 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 Regional Manager. For assistance, contact [Customer Service](#).

Test Classification

Not Applicable

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
THEV0	Thalassemia Summary Interpretation	14869-2

Result ID	Test Result Name	Result LOINC Value
608092	Thalassemia Summary Interpretation	14869-2
608118	Reviewed By	18771-6