Test Definition: THEV0

Thalassemia Summary Interpretation, Blood

Overview

Useful For

Incorporating and summarizing subsequent molecular results into an overall interpretation for the THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum

Testing Algorithm

When 1 or more molecular tests are added to the THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum, 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 THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum.

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

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 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 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 and Serum.

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 September 15, 2023. Accessed November 18, 2024. Available at www.omim.org/entry/141800?search=141800&highlight=141800
2. OMIM. 141900 Hemoglobin-beta locus; HBB. Updated September 15, 2023. Accessed November 18, 2024. Available at www.omim.org/entry/141900?search=141900&highlight=141900
3. 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
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. 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;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(23):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

Performing Laboratory Location

Mayo Clinic Laboratories - Rochester Main Campus

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
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