MAYO CLINIC LABORATORIES

Test Definition: THEVI

Hemoglobinopathy Interpretation

Overview

Useful For

Interpretation of results for the evaluation of thalassemias and hemoglobinopathies

Evaluation of microcytosis

Extensive and economical diagnosis and classification of hemoglobinopathies or thalassemia including complex disorders

Diagnosis of hereditary persistence of hemoglobin

Method Name

Only orderable as part of a profile. 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	7 days	

Clinical & Interpretive

Clinical Information

This consultative study is primarily designed for the evaluation of microcytosis but also can test for the detection of almost all known hemoglobin disorders in an economical manner. Because this can include multiple tests for alpha thalassemias, beta thalassemias, delta-beta thalassemia, hereditary persistence of fetal hemoglobin (HPFH), and for all known hemoglobin (Hb) variants, an expert in these disorders can guide testing to explain the clinical question or reported complete blood count values. This evaluation is particularly useful for complete classification of compound combinations of Hb S with alpha or beta thalassemia, Hb E/beta-0 thalassemia, and many other complex alpha and beta thalassemia disorders. Since iron deficiency can mimic thalassemias, if a serum sample is received, ferritin levels are

measured to evaluate this possibility.

Hb disorders include those associated with thalassemias (decreased protein quantity) and Hb variants (abnormal protein production). Many are clinically harmless, while others cause symptoms including microcytosis, sickling disorders, hemolysis, erythrocytosis, cyanosis/hypoxia, long-standing or familial anemia, compensated or episodic anemia, and increased methemoglobin or sulfhemoglobin results. Hb disorders can show patterns of either autosomal recessive or autosomal dominant inheritance.

The thalassemias are a group of disorders of Hb synthesis. Normal adult Hb consists of 2 alpha globin chains (encoded by 2 pairs of alpha globin genes, each pair located on chromosome 16), and 2 beta globin chains (encoded by 2 beta globin genes, each located on chromosome 11). Thalassemia syndromes result from an underproduction of 1 or 2 types of globin chains and are characterized by the type (alpha, beta, delta, gamma), magnitude of underproduction (number of defective genes), and the severity of clinical symptoms (minor, intermedia, major). The severity of the clinical and hematologic effects is directly related to the imbalance of alpha-like to beta-like chains.

The most common form of thalassemia is alpha thalassemia. Alpha thalassemia usually involves deletion of entire alpha genes and varies in severity depending on the number of alpha chains deleted (or rendered nonfunctional). Alpha thalassemia trait usually results from the deletion of 2 alpha genes. The most common form of Hb H disease results from dysfunction of 3 alpha chains and shows a variable phenotype, with most cases showing moderate anemia. The deletion of all 4 alpha genes (Barts hydrops fetalis) is incompatible with life without significant medical intervention.
Non-deletion alpha thalassemia genetic variants can also result in either thalassemia trait or Hb H disease and are less common than deletion forms.

Conversely most beta thalassemia genetic variants are due to single nucleotide substitutions that can occur anywhere in the beta globin gene. Large deletions of the beta globin gene complex can result in elevations in Hb F, such as HPFH or delta-beta thalassemia. While the presence of a single beta gene variant (beta thalassemia trait) results primarily in red blood cell microcytosis, cases with two beta gene abnormalities show a wide range in clinical severity, and many cases require molecular testing to understand the phenotype.

Reference Values

Only orderable as part of a profile. For more information see THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum.

Definitive results and an interpretive report will be provided.

Interpretation

A hematopathologist expert in these disorders evaluates the case and an interpretive report is issued.

Cautions

No significant cautionary statements

Clinical Reference

1. Hoyer JD, Hoffman DR. The thalassemia and hemoglobinopathy syndromes. In: McClatchey KD, Amin HM, Curry JL, eds. Clinical Laboratory Medicine. 2nd ed. Lippencott Williams and Wilkins; 2002:866-892
2. Brancaleoni V, Di Pierro E, Motta I, Cappellini MD. Laboratory diagnosis of thalassemia. Int J Lab Hematol. 2016;38 (Suppl 1):32-40

3. Hartveld C. State of the art and new developments in molecular diagnostics for hemoglobinopathies in multiethnic societies. Int J Lab Hematol. 2013;36:1-12

Performance

Method Description
A hematopathologist expert in these disorders evaluates the case, appropriate tests are performed, and an interpretive report is issued.

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
2 to 25 days if structural and/or molecular studies are required.

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

CPT Code Information
83020-26

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
THEVI	Hemoglobinopathy Interpretation	14869-2

Result ID	Test Result Name	Result LOINC® Value
608425	Hemoglobinopathy Interpretation	13514-5
608868	Reviewed By	18771-6