

Overview

Useful For

Interpretation for the results of hemoglobin electrophoresis

Diagnosis and classification of hemoglobin disorders, including thalassemias and hemoglobin variants

Special Instructions

- [Metabolic Hematology Patient Information](#)

Method Name

Only orderable as part of a profile. For more information see HBEL1 / Hemoglobin Electrophoresis 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 (preferred)	7 days	

Clinical & Interpretive

Clinical Information

A large number of variants of hemoglobin (Hb) have been recognized. Although many do not result in clinical or hematologic effects, clinical symptoms that can be associated with Hb disorders include microcytosis, sickling disorders, hemolysis, erythrocytosis/polycythemia, cyanosis/hypoxia, anemia (chronic, compensated or episodic), and increased methemoglobin or sulfhemoglobin results (M-Hb).

For many common Hb variants (eg, Hb S, Hb C, Hb D and Hb E, among many others), protein studies will be sufficient for definitive identification. However, some Hb conditions may be difficult to identify by protein methods alone and may require molecular methods for confirmation. Hb disorders commonly occur as compound disorders (2 or more genetic variants) that can have complex interactions and variable phenotypes. In these situations molecular testing may be necessary for accurate classification. It is important to note that although powerful as an adjunct for a complete and accurate diagnosis, molecular methods without protein data can give incomplete and possibly misleading information due to limitations of the methods. Accurate classification of Hb disorders and interpretation of genetic data requires the incorporation of protein analysis results. This profile is well-suited for the classification of Hb disorders.

Mayo Clinic Laboratories receives specimens from a wide geographic area and nearly one-half of all specimens tested exhibit abnormalities. The most common abnormality is an increase in Hb A2 to about 4% to 8%, which indicates beta-thalassemia minor when present in the correct clinical context. A wide variety of other hemoglobinopathies are also frequently encountered. Ranked in order of relative frequency, these are: Hb S (sickle cell disease and trait), C, E, Lepore, G-Philadelphia, Hb H disease, D-Los Angeles, Koln, Constant Spring, O-Arab. Other variants associated with hemolysis, erythrocytosis/polycythemia, microcytosis, cyanosis/hypoxia are routinely identified; however, some will not be detected by routine screening methods and require communication of clinical findings to prompt indicated reflex testing options. Alpha-thalassemia genetic variants are very common in the United States, occurring in approximately 30% of African Americans and accounting for the frequent occurrence of microcytosis in persons of this ethnic group. Some alpha-thalassemia conditions (eg, Hb H, Barts) can be identified in the Hb electrophoresis protocol, although Hb Constant Spring may or may not be evident by protein methods alone dependent upon the percentage present. It is important to note, alpha-thalassemias that are from only 1 or 2 alpha-globin gene deletions are not recognized by protein studies alone and alpha gene deletion and duplication testing is required.

Reference Values

Only orderable as part of a profile. For more information see HBEL1 / Hemoglobin Electrophoresis Evaluation, Blood.

Definitive results and an interpretative report will be provided.

Interpretation

Abnormal hemoglobin variants are identified. An interpretive report will be provided.

Cautions

Some hemoglobin disorders and variants are not detected by our screening methods, including common alpha thalassemia conditions, and require further reflex testing to identify. If a family history of a known hemoglobin disorder, prior therapy for a hemoglobin disorder, or otherwise unexplained lifelong/familial symptoms such as hemolysis, microcytosis, erythrocytosis/polycythemia, cyanosis, or hypoxia are present, this should be clearly communicated to the laboratory so appropriate reflex testing can be added, see [Metabolic Hematology Patient Information](#) in Special Instructions.

Recent transfusion may mask protein results including hemoglobin electrophoresis, hereditary persistence of fetal hemoglobin (HPFH) by flow cytometry, stability studies and sickle solubility studies depending on percentage of transfused cells present.

Some hemoglobin variants can originate from the donor blood product and not from the tested recipient. These are typically found in low percentage.

If the patient has undergone a bone marrow transplant, the results may show atypical results and should be interpreted in the context of clinical information.

Some therapies cause artefactual effects in protein studies, including hydroxyurea and decitabine (increased Hb F levels), Voxelotor (artefactual peaks) and gene therapy (alternate protein detection, Beta T87Q, by mass spectrometry). Clear communication of prior therapy is strongly recommended.

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. Lippincott Williams and Wilkins; 2002:866-895
2. Oliveira, JL: Diagnostic strategies in hemoglobinopathy testing, the role of a reference laboratory in the USA. Thalassemia Reports. 2018;8(1). doi: 10.4081/thal.2018.7476
3. Brancaleonai V, Di Pierro E, Motta I, Cappellini MD: Laboratory diagnosis of thalassemia. Int J Lab Haematol. 2016;38(suppl 1):32-40
4. Hartveld CI: State of the art and new developments in molecular diagnostics for hemoglobinopathies in multiethnic societies. Int J Lab Haematol. 2014;36:1-12
5. Szuberski J, Oliveira JL, Hoyer JD: A comprehensive analysis of hemoglobin variants by high-performance liquid chromatography (HPLC). Int J Lab Hematol. 2012;34(6):594-604
6. Riou J, Szuberski J, Godart C, et al: Precision of CAPILLARYS 2 for the Detection of Hemoglobin Variants Based on Their Migration Positions. Am J Clin Pathol. 2018;149(2):172-180

Performance

Method Description

A hematopathologist evaluates all of the testing performed and an interpretive report is provided.

PDF Report

No

Specimen Retention Time**Performing Laboratory Location**

Rochester

Fees & Codes**Test Classification**

Not Applicable

CPT Code Information

NA - Not billable

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
HBELI	Hb Electrophoresis Interpretation	In Process

Result ID	Reporting Name	LOINC®
608088	Hb Electrophoresis Interpretation	49316-3
609421	Hb Electrophoresis Interp Cancel	No LOINC Needed