



Reporting Title: Thalassemia and Hemoglobinopathy Ev

Performing Location: Rochester

Necessary Information:

Include the following information with the specimen:

Recent transfusion information

-Most recent complete blood cell count results

-If not sending serum, include ferritin results.

Metabolic Hematology Patient Information (T810) is strongly recommended. Testing may proceed without this information, however if the information requested is received, any pertinent reported clinical features and data will drive the focus of the evaluation and be considered in the interpretation.

The laboratory has extensive experience in hemoglobin variant identification and many cases can be confidently classified without molecular testing. However, molecular confirmation is always available, subject to sufficient sample quantity (eg, MLPA testing requires at least 2 mL of sample in addition to protein testing requirements). If no molecular testing or specific molecular tests are desired, utilize the appropriate check boxes on the form. If the form or other communication is not received, the reviewing hematopathologist will select appropriate tests to sufficiently explain the protein findings which may or may not include molecular testing.

Specimen Requirements:

Blood and serum are required.

Specimen Type: Whole blood

Container/Tube: Lavender top (EDTA)

Specimen Volume: 15 mL

Collection Instructions: Send whole blood specimen in original tube. Do not aliquot.

Specimen Type: Serum

Patient Preparation: For 12 hours before specimen collection do not take multivitamins or dietary supplements containing biotin (vitamin B7), which is commonly found in hair, skin, and nail supplements and multivitamins.

Collection Container/Tube:

Preferred: Serum gel

Acceptable: Red top

Submission Container/Tube: Plastic vial

Specimen Volume: 0.6 mL

Collection Instructions:

1. Serum gel tubes should be centrifuged within 2 hours of collection.
2. Red-top tubes should be centrifuged, and the serum aliquoted into a plastic vial within 2 hours of collection.
3. Label specimen as serum.

Specimen Minimum Volume:

Blood: 2.5 mL

Serum: 0.5 mL



Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
 - Informed Consent for Genetic Testing (T576)
 - Informed Consent for Genetic Testing-Spanish (T826)
2. Metabolic Hematology Patient Information (T810)
3. If not ordering electronically, complete, print, and send a Benign Hematology Test Request (T755) with the specimen

Specimen Type	Temperature	Time	Special Container
Serum	Refrigerated	7 days	
Whole Blood EDTA	Refrigerated	7 days	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
608425	Hemoglobinopathy Interpretation	Alphanumeric		13514-5
608868	Reviewed By	Alphanumeric		18771-6
41927	Hb A Also used by tests: HGBCE	Numeric	%	20572-4
41928	Hb F Also used by tests: HGBCE	Numeric	%	32682-7
41929	Hb A2 Also used by tests: HGBCE	Numeric	%	4552-6
41930	Variant 1 Also used by tests: HGBCE	Alphanumeric	%	24469-9
41931	Variant 2 Also used by tests: HGBCE	Alphanumeric	%	24469-9
41932	Variant 3 Also used by tests: HGBCE	Alphanumeric	%	24469-9
41933	HGBCE Interpretation Also used by tests: HGBCE	Alphanumeric		78748-1
65615	HPLC Hb Variant, B	Alphanumeric		No LOINC Needed



Result ID	Reporting Name	Type	Unit	LOINC®
FERR1	Ferritin, S Also used by tests: FERR1	Numeric	mcg/L	20567-4

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
THEV1	Hemoglobinopathy Interpretation			Yes	No
HGBCE	Hb Variant, A2 and F Quantitation,B			Yes	Yes
HPLC	HPLC Hb Variant, B			Yes	No
FERR1	Ferritin, S			Yes	Yes

CPT Code Information:

83020-26-Hemoglobinopathy Interpretation

83020-Hb Variant, A2 and F Quantitation

83021

82728

82664 (if appropriate)

83068 (if appropriate)

83789 (if appropriate)

88184 (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
HPFH	Hb F Distribution, B			No	No
SDEX	Sickle Solubility, B			No	Yes
IEF	Isoelectric Focusing, B			No	No



Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
UNHB	Hb Stability, B			No	No
MASS	Hb Variant by Mass Spec, B			No	No
ATHAL	Alpha-Globin Gene Analysis			No	Yes
WASQR	Alpha Globin Gene Sequencing, B			No	Yes (Order WASEQ)
WBSQR	Beta Globin Gene Sequencing, B			No	Yes (Order WBSEQ)
WBDDR	Beta Globin Cluster Locus Del/Dup,B			No	Yes (Order WBDD)
WGSQR	Gamma Globin Full Gene Sequencing			No	Yes (Order WGSEQ)
THEV0	Thalassemia Summary Interpretation			No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
HPFH	8270	Hb F Distribution, B	Alphanumeric		4579-9
HPFH	2104	Interpretation	Alphanumeric		59466-3
SDEX	9180	Sickle Solubility, B	Alphanumeric		6864-3
IEF	81644	Isoelectric Focusing, B	Alphanumeric		49316-3
UNHB	9095	Hb Stability, B	Alphanumeric		4639-1
MASS	60286	Hb Variant by Mass Spec, B	Alphanumeric		No LOINC Needed
ATHAL	52834	Result Summary	Alphanumeric		50397-9
ATHAL	52835	Result	Alphanumeric		82939-0
ATHAL	52836	Interpretation	Alphanumeric		69047-9
ATHAL	54871	Additional Information	Alphanumeric		48767-8
ATHAL	52837	Specimen	Alphanumeric		31208-2
ATHAL	52838	Source	Alphanumeric		31208-2
ATHAL	52839	Method	Alphanumeric		85069-3
ATHAL	52840	Released By	Alphanumeric		18771-6
WASQR	47952	Alpha Globin Gene Sequencing Result	Alphanumeric		50397-9



Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
WASQR	47953	Interpretation	Alphanumeric		59466-3
WBSQR	47954	Beta Globin Gene Sequencing Result	Alphanumeric		50397-9
WBSQR	47955	Interpretation	Alphanumeric		59466-3
WBDDR	48356	Beta Globin Cluster Locus Del/Dup	Alphanumeric		50397-9
WBDDR	48355	Reviewed by	Alphanumeric		18771-6
WBDDR	48357	Interpretation	Alphanumeric		59466-3
WGSQR	47950	Gamma Globin Gene Sequencing Result	Alphanumeric		50397-9
WGSQR	47951	Gamma Globin Interpretation	Alphanumeric		59466-3
THEV0	608092	Thalassemia Summary Interpretation	Alphanumeric		14869-2
THEV0	608118	Reviewed By	Alphanumeric		18771-6

Reference Values:

Definitive results and an interpretive report will be provided.