



Test Definition: THEV1

Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum

Reporting Title: Thalassemia and Hemoglobinopathy Ev

Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

Ordering Guidance:

Multiple hematology evaluations are available. For information on testing that is performed with each evaluation, see [Benign Hematology Evaluation Comparison](#).

If monitoring treatment with HbA-T87Q (Lyfgenia for sickle cell disorders or Zynteglo for thalassemia), complete [Metabolic Hematology Patient Information](#) (T810) to notify the laboratory and request mass spectrometry testing.

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 hour before specimen collection, patient **should not** take multivitamins or dietary supplements (eg, hair, skin, and nail supplements) containing biotin (vitamin B7).

Collection Container/Tube:

Preferred: Serum gel

Acceptable: Red top

Submission Container/Tube: Plastic vial

Specimen Volume: 0.6 mL

Collection Instructions:

1. Within 2 hours of collection, serum gel tubes should be centrifuged.

2. Within 2 hours of collection, red-top tubes should be centrifuged and the serum aliquoted into a plastic vial.
3. Label specimen as serum.

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®
41927	Hb A	Numeric	%	20572-4
41928	Hb F	Numeric	%	32682-7
41929	Hb A2	Numeric	%	4552-6
41930	Variant 1	Alphanumeric	%	24469-9
41931	Variant 2	Alphanumeric	%	24469-9
41932	Variant 3	Alphanumeric	%	24469-9
41933	HGBCE Interpretation	Alphanumeric		78748-1
FERR1	Ferritin, S	Numeric	mcg/L	20567-4
65615	HPLC Hb Variant, B	Alphanumeric		No LOINC Needed
608425	Hemoglobinopathy Interpretation	Alphanumeric		13514-5
608868	Reviewed By	Alphanumeric		18771-6

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

Components:

Test Id	Reporting Name	CPT Units	CPT Code	Always Performed	Available Separately
THEVI	Hemoglobinopathy Interpretation	1	83020	Yes	No
HGBCE	Hb Variant, A2 and F Quantitation,B	1	83020	Yes	Yes
HPLC	HPLC Hb Variant, B	1	83021	Yes	No
FERR1	Ferritin, S	1	82728	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	Available Separately
HPFH	Hb F Distribution, B	1	88184	No	No
SDEX	Sickle Solubility, B	1	85660	No	Yes
IEF	Isoelectric Focusing, B	1	82664	No	No
UNHB	Hb Stability, B	1	83068	No	No
MASS	Hb Variant by Mass Spec, B	1	83789	No	No
WASQR	Alpha Globin Gene Sequencing, B	1	81259	No	Yes, (Order WASEQ)
WBSQR	Beta Globin Gene Sequencing, B	1	81364	No	Yes, (Order WBSEQ)
WGSQR	Gamma Globin Full Gene Sequencing	1	81479	No	Yes, (Order WGSEQ)
THEV0	Thalassemia Summary Interpretation			No	No
WAGDR	Alpha Globin Clustr Locus Del/Dup,B	1	81269	No	Yes, (Order AGDD)
WBGDR	Beta Globin Gene Cluster, Del/Dup,B	1	81363	No	Yes, (Order WBGDD)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
IEF	81644	Isoelectric Focusing, B	Alphanumeric		49316-3
SDEX	9180	Sickle Solubility, B	Alphanumeric		6864-3
MASS	60286	Hb Variant by Mass Spec, B	Alphanumeric		No LOINC Needed
HPFH	8270	Hb F Distribution, B	Alphanumeric		4579-9
HPFH	2104	Interpretation	Alphanumeric		59466-3
UNHB	9095	Hb Stability, B	Alphanumeric		4639-1
WASQR	47952	Alpha Globin Gene Sequencing Result	Alphanumeric		50397-9
WASQR	47953	Interpretation	Alphanumeric		59466-3
WBSQR	47954	Beta Globin Gene Sequencing Result	Alphanumeric		50397-9
WBSQR	47955	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

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Thalassemia and Hemoglobinopathy
Evaluation, Blood and Serum

THEV0	608118	Reviewed By	Alphanumeric		18771-6
WBGDR	620977	Beta Globin Gene Cluster Del/Dup	Alphanumeric		101634-4
WBGDR	620978	Reviewed by	Alphanumeric		18771-6
WBGDR	620976	Interpretation	Alphanumeric		69047-9
WAGDR	621362	Result Summary	Alphanumeric		50397-9
WAGDR	621363	Result	Alphanumeric		82939-0
WAGDR	621364	Interpretation	Alphanumeric		69047-9
WAGDR	621365	Additional Information	Alphanumeric		48767-8
WAGDR	621366	Specimen	Alphanumeric		31208-2
WAGDR	621367	Source	Alphanumeric		31208-2
WAGDR	621368	Method	Alphanumeric		85069-3
WAGDR	621804	Disclaimer	Alphanumeric		62364-5
WAGDR	621369	Released By	Alphanumeric		18771-6

Reference Values:

Definitive results and an interpretive report will be provided.