
Reporting Title: Alpha Globin Gene Sequencing, B**Performing Location:** Rochester**Ordering Guidance:**

For first-tier testing for alpha thalassemia detection, order THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Serum and Whole Blood.

For first-tier testing for an alpha-globin variant, order HBEL1 / Hemoglobin Electrophoresis Evaluation, Blood.

If genetic testing is desired, the first-tier genetic test assesses large deletional alpha-thalassemia alterations. Order ATHAL / Alpha-Globin Gene Analysis, Varies.

Necessary Information:

1. Patient's age is required.
2. Include recent transfusion information.

Specimen Requirements:**Submit only 1 of the following specimens:****Specimen Type:** Whole blood**Container/Tube:****Preferred:** Lavender top (EDTA)**Acceptable:** Yellow top (ACD), green top (sodium heparin)**Specimen Volume:** 4 mL**Collection Instructions:**

1. Invert several times to mix blood.
2. Send whole blood specimen in the original tube. **Do not aliquot**

Specimen Stability Information: Refrigerate 30 days(preferred)/Ambient 14 days**Specimen Type:** Extracted DNA from whole blood**Container/Tube:** 1.5- to 2-mL tube**Specimen Volume:** Entire specimen**Collection Instructions:**

1. Label specimen as extracted DNA and source of specimen
2. Provide volume and concentration of the DNA

Specimen Stability Information: Frozen (preferred)/Refrigerate/Ambient**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 in Special Instructions:

-[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 [Benign Hematology Test Request Form](#) (T755) with the specimen

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
43921	Interpretation	Alphanumeric		69047-9
61362	Alpha Globin Gene Sequencing, B	Alphanumeric		87730-8

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

Supplemental Report:

No

CPT Code Information:

81259-HBA1/HBA2; full sequence

Reference Values:

An interpretive report will be provided.