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

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

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

Necessary Information:

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

Specimen Requirements:

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

Specimen Minimum Volume:

Blood: 1 mL; Extracted DNA: 50 mcL at 50 ng/mcL concentration

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
Varies	Varies		

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
62128	Beta Globin Gene Sequencing, B	Alphanumeric		82939-0
43922	Interpretation	Alphanumeric		69047-9

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81364-HBB (hemoglobin, beta) full sequence

Reference Values:

An interpretive report will be provided.