Test Definition: WBSEQ

Beta Globin Gene Sequencing, Varies

Reporting Title: Beta Globin Gene Sequencing, B
Performing Location: Mayo Clinic Laboratories - Rochester Main Campus

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 whole 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. DNA must be extracted from whole blood within 7 days of collection.
2. Label specimen as extracted DNA and source of specimen.
3. Provide volume and concentration of the DNA.
Specimen Stability Information: Frozen (preferred) 1 year/Refrigerate/Ambient
Additional Information: DNA must be extracted in a CLIA-certified laboratory or equivalent and must be extracted from a specimen type listed as acceptable for this test (including applicable anticoagulants). We cannot guarantee that all extraction methods are compatible with this test. If testing fails, one repeat will be attempted, and if unsuccessful, the test will be reported as failed and a charge will be applied.

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.

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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.