

## **ALPHA GLOBIN GENE SEQUENCING, B**

### **Test ID: WASEQ**

**TESTING ALGORITHM:**

This is a second-tier test in the evaluation of alpha thalassemia carrier determination, hemoglobin H disease confirmation, and alpha globin variant identification.

First-tier testing for alpha thalassemia detection is THEVP / Thalassemia and Hemoglobinopathy Evaluation. If only genetic testing is desired, the first-tier genetic test is for large deletional alpha thalassemia mutations (ATHAL / Alpha-Globin Gene Analysis). First-tier testing for an alpha globin variant is HBELC / Hemoglobin Electrophoresis Cascade, Blood.

**USEFUL FOR:**

- Diagnosing nondeletional alpha thalassemia.
- Testing for nondeletional alpha thalassemia in a symptomatic individual.
- Follow-up testing to an abnormal hemoglobin electrophoresis that identified an alpha globin chain variant.

**METHOD:** Polymerase Chain Reaction (PCR) followed by DNA Sequence Analysis

**REFERENCE VALUES:** An interpretive report will be provided.

**SPECIMEN REQUIREMENTS:****Container/Tube:**

**Preferred:** EDTA

**Acceptable:** ACD, Sodium Heparin

**Specimen Volume:** 4mL

**Minimum Volume:** 1mL

**Collection Instructions:** Do not transfer blood to other containers

**SPECIMEN STABILITY INFORMATION:**

| Specimen Type    | Temperature  | Time    |
|------------------|--------------|---------|
| Whole Blood EDTA | Refrigerated | 14 days |

**CAUTIONS:**

This assay will not detect large deletions or duplications within the alpha globin genes. Therefore, test results should be interpreted in the context of hemoglobin electrophoresis, clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

**CPT CODE:** 81257-HBA1/HBA2 (alpha globin 1 and alpha globin 2)

**DAY(S) SET UP:** Monday/Wednesday/Friday **ANALYTIC TIME:** 2 days

NOTE: The following referral test code(s) will become obsolete.

| Test Name   | Test ID | Referral Lab Code | Referral Lab      |
|---|---------|-------------------|-------------------|
| Alpha Globin (HBA1 and HBA2)<br>Sequencing                          | ZW242   | 2001582           | ARUP Laboratories |
| Alpha Globin (HBA1 and HBA2)<br>Sequencing and Deletion/Duplication | ZW242   | 2011708           | ARUP Laboratories |

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or  
Melissa Lonzo, MML Laboratory Technologist Resource Coordinator  
Telephone: 800-533-1710