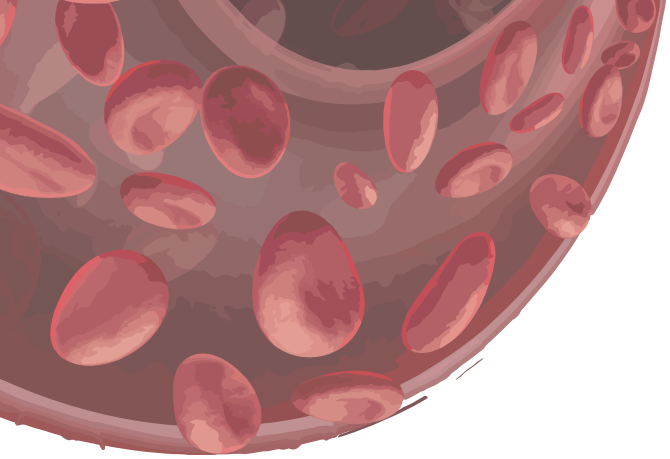

**Hemoglobinopathies are the most common monogenic diseases.** ARUP has a comprehensive offering of hemoglobinopathy testing, from HPLC to genetic confirmation. Hemoglobin testing is particularly indicated in the following situations:

- Microcytic hypochromic anemia after iron deficiency has been ruled out
- Chronic hemolytic anemia
- Vascular obliteration crises of unclear etiology in patients from areas where HbS and/or HbC is widespread
- Drug-induced anemia
- Erythrocytosis and/or cyanosis caused by hematological factors
- Hydrops fetalis of unclear etiology
- Prevention (e.g., testing family members or partners)
- Prenatal diagnosis

**www.aruplab.com/topics/hemoglobinopathy**

## Reference

Kohne E. Hemoglobinopathies. *Dtsch Arztebl Int* 2011;108(31–32):532–40.

ARUP LABORATORIES

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keyword: Hemoglobinopathy

*A nonprofit enterprise of the University of Utah and its Department of Pathology*


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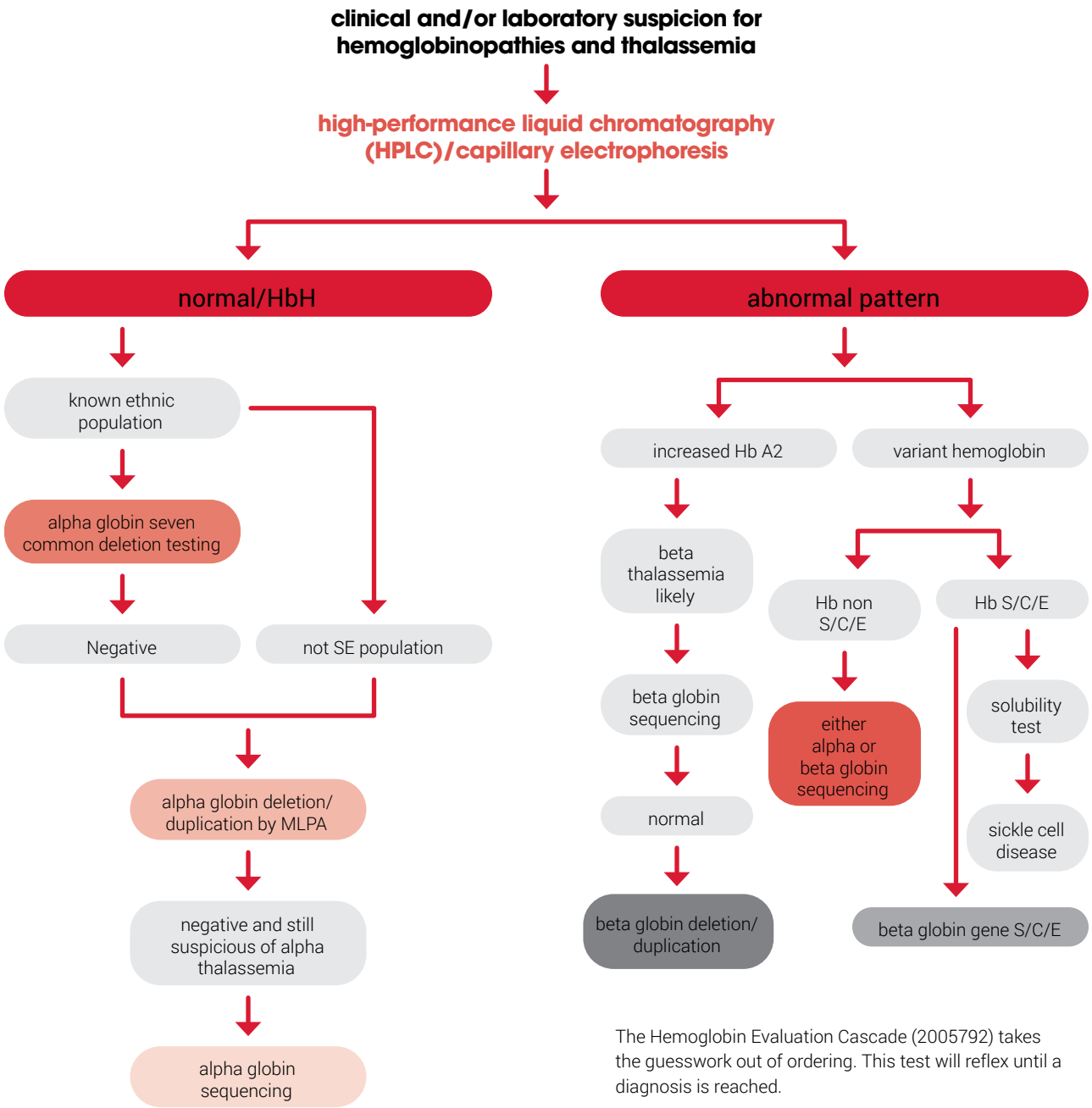
# Hemoglobinopathy

testing at ARUP Laboratories

ARUP LABORATORIES

# Hemoglobinopathy Testing Algorithm




The Hemoglobin Evaluation Cascade (2005792) takes the guesswork out of ordering. This test will reflex until a diagnosis is reached.

For more information and educational opportunities, visit:
**www.aruplab.com/topics/hemoglobinopathy**

# ARUP Hemoglobinpathy Test Menu

**Hemoglobin Evaluation Reflexive Cascade (2005792)**
Optimal test for initial and confirmatory diagnosis of any suspected hemoglobinopathy.

**Hemoglobin Evaluation with Reflex to Electrophoresis and/or RBC Solubility (0050610)**
Effective test for screening and follow-up of individuals with known hemoglobinopathies.

**Hemoglobin S, Evaluation with Reflex to RBC Solubility (0050520)**
Screens for presence of hemoglobin S; does not differentiate between sickle cell disease (homzygous S/S) and sickle cell trait (carrier).

**Hemoglobin S, Sickle Solubility (2013399)**
Effective test for secondary confirmation of hemoglobin (Hb) S. Not recommended for newborns <6 months due to high concentration of HbF.

**Hemoglobin (Hb) A[2] and F by Column (0050613)**
Quantifies HbA2 and HbF in whole blood; aids in the management of sickle cell disease and in the identification of beta thalassemia carriers.

**Hemoglobin F (0081348)**
Measures percentage of hemoglobin F only.

**Alpha Globin (*HBA1* and *HBA2*) Deletion/Duplication (2011622)**
Preferred first-tier genetic test for confirmation of suspected alpha thalassemia or alpha thalassemia trait; detects common, rare, and novel deletions or duplications of the alpha globin gene cluster.

**Alpha Thalassemia (*HBA1* and *HBA2*) 7 Deletions (0051495)**
Acceptable first-tier genetic test for confirmation of suspected alpha thalassemia or alpha thalassemia trait; assesses for seven common alpha globin gene deletions.

**Alpha Globin (*HBA1* and *HBA2*) Sequencing and Deletion/Duplication (2011708 )**
Comprehensive test for detection of alpha thalassemia or alpha thalassemia trait.

**Alpha Globin (*HBA1* and *HBA2*) Sequencing (2001582)**
Commonly used as a second-tier test for detection of alpha thalassemia; requires approval from an ARUP genetic counselor.

**Beta Globin (HBB) Sequencing and Deletion/Duplication (2010117)**
Preferred test for molecular confirmation of beta thalassemia or a hemoglobinopathy involving the beta-globin gene.

**Beta Globin (HBB) Gene Sequencing (0050578)**
Molecular confirmation of a suspected structural hemoglobinopathy or beta thalassemia.

**Beta Globin (HBB) Deletion/Duplication (2010113)**
Second-tier test; requires approval from an ARUP genetic counselor.

**Beta Globin (HBB) HbS, HbC, and HbE Mutations (0051421)**
Confirms suspected HbS, HbC, and HbE mutations.

**Beta Globin (HBB) HbS, HbC, and HbE Mutations, Fetal (0051422)**
Genetic test on fetal samples for prenatal detection of HbS, HbC, and HbE mutations.

**Familial Mutation, Targeted Sequencing (2001961)**
Detects mutation previously identified in a family member; consultation with an ARUP genetic counselor is advised.

**Familial Mutation, Targeted Sequencing, Fetal (2001980)**
Fetal testing to detect a previously characterized mutation in a family member; requires approval from an ARUP genetic counselor.