

Client: Example Client ABC123  
123 Test Drive  
Salt Lake City, UT 84108  
UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 1/2/1945  
**Gender:** Male  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Hemoglobin Lepore (HBD/HBB Fusion) 3 Mutations**

ARUP test code 2004686

Hemoglobin Lepore 3 Mutations Specimen whole Blood

Hemoglobin Lepore 3 Mutations

Het WB

Indication for testing: Confirm presence of hemoglobin Lepore.

Result: Heterozyous Hb Lepore-Washington-Boston  
Mutation: g.63632\_71046del

Interpretation: One copy of the delta/beta globin fusion gene, Hb Lepore-Washington-Boston, was detected. This individual is at least a carrier of Hb Lepore; carriers typically present with mild anemia, hypochromic microcytosis, and moderately increased fetal hemoglobin. Individuals who are compound heterozygotes for Hb Lepore and a second beta globin gene mutation (not identified by this assay) have variable clinical presentations.

Recommendations: Medical management should rely on clinical findings. Testing for hemoglobin variants (Hemoglobin Evaluation; ARUP test #0050610) should be offered to this individual's reproductive partner and family members to assess their carrier status. A genetics consultation is recommended.

Reference sequence: NG\_000007.3

This result has been reviewed and approved by Wei Shen, Ph.D.

H=High, L=Low, \*=Abnormal, C=Critical

**BACKGROUND INFORMATION: Hemoglobin Lepore (HBD/HBB Fusion)  
 3 Mutations**

**CHARACTERISTICS:** Hb Lepore is a hemoglobin variant resulting from a fusion between the delta globin gene (HBD) and the beta globin gene (HBB). Hb Lepore is classified as a beta-plus thalassemia mutation, as it results in reduced beta globin chain synthesis and in its heterozygous form is associated with mild anemia, hypochromic microcytosis, and moderately increased fetal hemoglobin.  
**INCIDENCE:** Most common in southern Europeans.  
**INHERITANCE:** Autosomal recessive.  
**CAUSE:** Delta/beta globin gene rearrangements.  
**MUTATIONS TESTED:** Hb Lepore-washington-Boston (g.63632\_71046del), Hb Lepore-Baltimore (g.63564\_70978del), and Hb Lepore-Hollandia (g.63290\_70702del).  
**CLINICAL SENSITIVITY:** Unknown.  
**METHODOLOGY:** Multiplex PCR and gel electrophoresis to detect the three common Hb Lepore mutations.  
**ANALYTICAL SENSITIVITY AND SPECIFICITY:** 99 percent.  
**LIMITATIONS:** Only the three common Hb Lepore mutations will be detected. Diagnostic errors can occur due to rare sequence variations.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

**VERIFIED/REPORTED DATES**

Procedure	Accession	Collected	Received	Verified/Reported
Hemoglobin Lepore 3 Mutations Specimen	18-333-107353	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin Lepore 3 Mutations	18-333-107353	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

**END OF CHART**

**H=High, L=Low, \*=Abnormal, C=Critical**