

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 10/2/2016
Gender: Female
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

Negative

Indication for testing: Confirm presence of hemoglobin Lepore.

Result: Negative

Interpretation: None of the three targeted Hb Lepore delta/beta globin gene mutations were detected. This result does not exclude thalassemia trait or disease as this individual may have other beta, delta, or alpha globin gene mutation(s) not identified by this assay.

Recommendation: Medical management should rely on clinical findings.

This result has been reviewed and approved by Rong Mao, M.D.

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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Hemoglobin Lepore 3 Mutations Specimen	18-165-402228	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin Lepore 3 Mutations	18-165-402228	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