Alpha Thalassemia (HBA1 and HBA2) 7 Deletions
ARUP test code 0051495

Alpha Thalassemia, Specimen
Whole Blood

Alpha Thalassemia, 7 Deletions
Negative

Indication for testing: Carrier screening or diagnostic testing for alpha thalassemia.

Negative: None of the common alpha globin gene deletions tested were detected. This reduces, but does not eliminate, the possibility of alpha-thalassemia as this test does not detect all alpha globin mutations.

This result has been reviewed and approved by Rong Mao, M.D.
BACKGROUND INFORMATION: Alpha Thalassemia (HBA1 and HBA2), 7 Deletions

CHARACTERISTICS:
- Alpha (+) thalassemia (silent carrier): Mutation of a single alpha2 globin gene (-a/aa); asymptomatic.
- Alpha (0) thalassemia (trait): Mutation of both alpha2 globin genes, or deletion of alpha 1 and alpha 2 globin genes in cis (-a/-a; --/aa); mild microcytic anemia possible.
- Hemoglobin H disease: Mutation of three alpha globin genes (--/-a); hemolysis with Heinz bodies, moderate anemia, splenomegaly.
- Hb Bart Hydrops Fetalis Syndrome: Mutation of four alpha globin genes (--/--); lethal in fetal or early neonatal period.

INCIDENCE: Carrier frequency in Mediterranean (1:30-50), Middle Eastern, Southeast Asian (1:20), African, African-American (1:3).

INHERITANCE: Autosomal recessive.

CAUSE: Mutations in the alpha globin gene cluster; 95 percent are deletions.

MUTATIONS TESTED: -a3.7, -a4.2, -(a)20.5, --SEA, --MED, --FIL, --THAI.

CLINICAL SENSITIVITY: Varies by ethnicity, may be as high as 90 percent.

METHODOLOGY: Polymerase chain reaction and gel electrophoresis.

ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.

LIMITATIONS: Rare alpha globin gene deletions, non-deletion mutations, gene duplications and mutations of the regulatory region will not 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

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Accession</th>
<th>Collected</th>
<th>Received</th>
<th>Verified/Reported</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alpha Thalassemia, Specimen</td>
<td>19-229-401341</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
</tr>
<tr>
<td>Alpha Thalassemia, 7 Deletions</td>
<td>19-229-401341</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
<td>00/00/0000 00:00</td>
</tr>
</tbody>
</table>

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