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

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

The 3.7 kb alpha globin gene deletion was detected and no normal alpha2 globin gene was present, thus, predicting the deletion of the alpha2 globin gene from both chromosomes. This result is consistent with alpha (0) thalassemia (trait) which is often associated with mild anemia and microcytosis. Carrier screening should be offered to the patient's reproductive partner and family members. Since this test does not detect all alpha globin mutations, additional testing should be considered if clinical symptoms are not consistent with the above diagnosis. This test is unable to differentiate homozygotes for the 3.7 kb deletion from compound heterozygotes for the 3.7 kb deletion and a second rare mutation.

This result has been reviewed and approved by Yuan Ji, Ph.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

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