Warde Medical Laboratory

LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

EXAMPLE, REPORT W
WX0000003827 M 07/08/1978 45 Y

Referral Testing

Collected: 08/17/2023 14:45 Received: 08/17/2023 14:45

Test Name	Result	Flag	Ref-Ranges	Units	Site
Alpha-Globin Common Mutation Analysis	**See Below**	**AB**			QCRL

RESULT: HETEROZYGOUS POSITIVE FOR THE -alpha3.7
ALPHA(PLUS)-THALASSEMIA MUTATION

Interpretation: DNA testing indicates that this patient is positive for the -alpha3.7 alpha-globin deletion on one chromosome. This deletion removes one of the alpha-globin genes from the alpha-globin gene cluster. Therefore, this patient is at least a carrier of an alpha(plus)-thalassemia mutation (genotype -alpha/alphaalpha).

Individuals with this genotype are usually clinically normal. If this patient is symptomatic, he or she may have an additional, rare alpha-thalassemia mutation. If the partner of this patient is a carrier of alpha(zero)-thalassemia, this couple is at-risk of having a child affected by Hemoglobin H disease. Family studies may be indicated. Genetic counseling is recommended.

Alpha-globin is an essential component of the hemoglobin tetramer, starting from the early stages of embryonic development. Deletion mutations involving one or both of the two alpha-globin genes (alpha1 and alpha2, located on chromosome 16p13) lead to reduced production of alpha-globin chains, and are the major cause of alpha-thalassemia. Severity of the disease is dependent on the total copy number of functional alpha-globin genes remaining.

This assay detects the seven most common deletions (-alpha3.7, -alpha4.2, -alpha20.5, --SEA, --MED, --FIL, and --THAI) found in patients with alpha-thalassemia. This assay is performed by allele-specific PCR amplification of deletion mutation fragments, followed by agarose gel electrophoresis of the amplification products. It is not known what percentage of individuals with alpha-globin gene deletions will be detected by this test.

For assistance with the interpretation of those results, please contact your local Quest Diagnostics genetic counselor or call 1-866-GENEINFO (436-3463).

This test is performed pursuant to a license agreement with Roche Molecular Systems, Inc. This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics Nichols Institute San Juan Capistrano. It has not been cleared or approved by the FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Test Performed at:

LAB: L - LOW, H - HIGH, AB - ABNORMAL, C - CRITICAL, . - NOT TESTED

F217000142
WX0000003827
Printed D&T: 08/17/23 14:48

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002354

Kajal V. Sitwala, MD, PhD - Medical Director
Form: MM RL1

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Quest Diagnostics Nichols Institute 33608 Ortega Highway San Juan Capistrano, CA 92675-2042	I Maramica MD, PhD, MBA				

Performing Site:
QCRL: QUEST DIAGNOSTICS REFERENCE LAB CAPISTRANO 33608 Ortega Highway San Juan Capistrano CA 92675

Reported Date: 2023.08.17 14:48

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