Warde Medical Laboratory

# LABORATORY REPORT

Example Client, XYZ123
1234 Warde Road
Ann Arbor MI 48108

**EXAMPLE, REPORT W**
WX0000003826 F 12/05/1988 34 Y

## Referral Testing

Collected: 08/31/2023 08:03 Received: 08/31/2023 08:03

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

RESULT: NO MUTATION DETECTED

Interpretation: DNA testing indicates that this patient is negative for the seven alpha-globin deletions (see below) commonly associated with alpha-thalassemia. This result does not rule out the presence of other mutations in the alpha-globin gene cluster. The residual risk for this patient to be a carrier of an alpha-thalassemia mutation is currently unknown.

Laboratory testing supervised and results monitored by Tina Hambuch-Hawks, Ph.D., DABMGG, CGMB.

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.
Reviewed and signed by Laboratory testing supervised and results monitored by Tina Hambuch-Hawks, Ph.D., DABMGG, CGMB, Signed on 08/14/2023 at 17:31
Test Performed at:
Quest Diagnostics Nichols Institute

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|---|---|---|---|---|---|
| 33608 Ortega Highway<br>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:** 2023.08.31 8:03

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

F231000001
WX0000003826
Printed D&T: 08/31/23 08:04

Ordered By: KAJAL SITWALA, MD, PhD
WX00000000002353

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