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: 08/31/2023 08:06 Received: 08/31/2023 08:06

Test Name	Result	Flag	Ref-Ranges	Units	Site
Alpha-globin Gene Del or Dup					
Alpha-globin Gene Del or Dup	**See Below**	**AB**			QCRL

RESULT: DELETION OF TWO ALPHA-GLOBIN GENES (2 ALPHA-GLOBIN GENES DETECTED)

INTERPRETATION: DNA testing indicates that this individual has two intact alpha-globin genes. The results are consistent with the presence of two copies of a fusion gene that consists of alpha-2 globin gene sequences at the 5'-end and alpha-1 globin gene sequences in the middle section and at the 3'-end. These results are consistent with the presence of two copies of the -alpha3.7 deletion. Additional testing (Test Code 11175, Alpha-Globin Common Mutation Analysis) can be performed to confirm the presence of the -alpha3.7 deletion.

Individuals with this genotype may have very mild anemia and hematological abnormalities including microcytosis and a variable degree of hypochromia. These individuals are at risk for having offspring with Hb H disease or alpha-thalassemia minor if their partners are alpha-thalassemia carriers. Testing of other family members may be appropriate. Genetic counseling is recommended.

This test cannot rule out the possibility of a point mutation in the alpha-globin gene cluster. If clinical and other laboratory findings are inconsistent with this result, consider alpha-globin gene sequencing.

Laboratory results and submitted clinical information reviewed by Carole Oddoux, Ph.D., DABMGG, CGMBS.

Alpha-globin is an essential component of the hemoglobin tetramer, beginning in the early stages of embryonic development. Deletion mutations involving one or both of the alpha-globin genes (designated HBA1 (alpha-1) and HBA2 (alpha-2)) in the alpha-globin gene cluster (located on chromosome 16p13) lead to reduced production of alpha-globin chains, and are the major cause of alpha-thalassemia. The severity of symptoms is dependent on the number of functional alpha-globin genes remaining.

In this assay, multiplex-polymerase chain reaction (PCR) amplification of fragments specific for the 5'-end, middle section, and 3'-end of the alpha1- and alpha2-globin genes is carried out in the presence of fluorescently-labeled primers. In addition, fragments representing the HS40/locus control region (LCR), the alpha-globin pseudogene, as well 3 non-alpha globin fragments used as internal

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controls, are co-amplified. Amplification products are analyzed on an automated capillary DNA sequencer.

This assay cannot detect single nucleotide substitutions and small insertion/deletion mutations in the alpha-globin gene cluster. Since genetic variation and other factors can affect the accuracy of direct mutation testing, the results of this testing should always be interpreted in light of clinical and familial data. For assistance with the interpretation of the results, please contact your local Quest Diagnostics genetic counselor or call 1-866-GENEINFO (436-3463).

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 FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.
Reviewed and signed by Laboratory results and submitted clinical information reviewed by Carole Oddoux, Ph.D., DABMGG, CGMBS, Signed on 08/15/2023 at 04:15
Test Performed at:
Quest Diagnostics Nichols Institute
33608 Ortega Highway
San Juan Capistrano, CA 92675-2042 I Maramica MD, PhD, MBA

Ethnicity:	NA	QCRL
Clinical Indication:	NA	QCRL
Referring Physician:	NA	QCRL
Physician Phone:	NA	QCRL

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

Reported Date: 2023.08.31 8:07 AGDEL