Patient ID	Patient Name		Birth Date	Gender	Age
SA00149284	VALIDATIONTESTING, THEV1		1978-08-08	F	43
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00149284	SA00149284	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		03 Nov 2021 14:00			

Alpha-Globin Gene Analysis

Alpha-Globin Gene Analysis (ATHL)

Result Summary

MCR

SILENT ALPHA THALASSEMIA CARRIER

Result

MCR

A single alpha globin gene is deleted (-3.7 Kb, "rightward") on one chromosome.

Interpretation

MCR

This result indicates that this individual is a silent alpha thalassemia carrier. While a silent carrier is not expected to develop symptoms of alpha thalassemia, mild microcytosis may be present. If appropriate, molecular testing on this individual's reproductive partner are recommended to further clarify their reproductive risk.

This assay does NOT detect non-deletion types of mutations (e.g., Constant Spring, alphaT Saudi). Therefore, this result should be interpreted in the context of clinical presentation and results of other laboratory tests [e.g. hemoglobin electrophoresis and mean corpuscular volume (MCV)].

A genetic consultation may be of benefit.

Specimen

MCR

WB Whole Blood

Method

MCR

Dosage analysis (PCR and MLPA) was used to detect deletion and duplication-type mutations. This method uses multiple probes that hybridize throughout the alpha-gene locus on chromosome 16 from the HS-40 regulatory region through the 3' hypervariable region (3'HVR).

Released By

MCR

JENNIFER MAIN

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects de-identified genetic and health information to advance the knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

One or more in silico tools were used to assist in the interpretation of these results. These tools are updated regularly and predictions for a given variant may change. Additionally, the predictability of these tools for the determination of pathogenicity is currently unvalidated.

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Received: 04 Nov 2021 09:24 **Reported:** 05 Nov 2021 09:33

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

Patient ID	Patient Name		Birth Date	Gender	Age
SA00149284	VALIDATIONTESTING, THEV1		1978-08-08	F	43
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
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C7028846 DLMP Rochester		03 Nov 2021 14:00			

Thalassemia Summary Interpretation

Thalassemia Summary Interpretation

MCR

Alpha Globin Cluster Del/Dup: Positive

The following alteration was detected:

Alpha Globin Gene Cluster:
DNA Change: Deletion,–3.7 Kb, rightward
Affected Locus: HBA2
Heterozygous
Classification: Alpha thalassemia mutation

SUMMARY INTERPRETATION:

1) The results indicate the functional loss of one alpha globin gene due to a heterozygous -3.7 kb deletion (-a/aa). A single deletion is associated with a clinically "silent carrier" phenotype but sometimes causes mild microcytosis. It is not associated with significant anemia. Clinical correlation is necessary. The -3.7 kb deletion is one of the most common alpha thalassemia mutations. It is often seen in people with African, South East Asian, Indian, Middle Eastern and Mediterranean heritage. (Harteveld 2010 PMID 20507641, Kipp 2011 PMID: 21708285)

2) Ferritin level is not supportive of iron deficiency. Correlation with iron studies is recommended.

3) No evidence of hemoglobin variants or beta thalassemia was detected by protein analysis. The vast majority of hemoglobin variants and beta thalassemias are excluded, although some rare clinically significant hemoglobin disorders are electrophoretically silent. Beta globin gene sequencing and beta globin cluster locus deletion/duplication analysis are available if clinically indicated. If desired, please call the Metabolic Hematology Laboratory at 1–800–533–1710.

GENETIC COUNSELING INFORMATION:

These results may have relevance for this individual's relatives or descendants. Single alpha globin gene deletions are typically clinically harmless. They can carry some reproductive risk or have some protective effect on the carrier's potential offspring. A single alpha globin deletion can carry some reproductive risk for Hb H disease if offspring co-inherits a cis alpha thalassemia deletion or another clinically significant alpha globin variant. Alpha globin gene deletions may also be protective, as they reduce co-inherited abnormal beta variant levels and balance alpha/beta chain ratios in the context beta thalassemia to modulate clinical severity.

Caution:
The alpha globin deletion/duplication assay (ATHAL) does not detect non-deletional types of alpha thalassemia mutations. DNA sequencing of the alpha globin genes is available if clinically indicated.

Reviewed By

MCR

JENNIFER MAIN

Received: 04 Nov 2021 09:24 **Reported:** 05 Nov 2021 10:47

Thalassemia and Hemoglobinopathy Ev

Hemoglobinopathy Interpretation

MCR

EVAL INCOMPLETE UNTIL SUMMARY INTERP ISSUED

Molecular testing is added. A summary interpretation with correlation of protein and molecular results will be reported when all testing under the profile is complete. Time to issuance of the final report is dependent on the complexity of the case. If the Thalassemia Summary Interpretation (Test ID THEV0) is not viewable 30 days after receipt of this protein interpretation, please call Mayo Clinic Laboratories to inquire and request a copy of the full report at 1–800–533–1710.

1) No abnormal hemoglobin variant or beta thalassemia detected by protein methods. Hb A2 is slightly decreased of uncertain

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Account Information		**Collected**			
C7028846 DLMP Rochester		03 Nov 2021 14:00			

significance.

2) The ferritin level is not supportive of iron deficiency. Correlation with iron studies is recommended.

Methodologies utilized in this interpretation include: capillary electrophoresis, HPLC

Reviewed By MCR

JENNIFER MAIN

Hb Variant, A2 and F Quantitation,B

Result Name	Value	Unit	Reference Value	Performing Site
⚠ High **Hb A**	**98.1**	%	95.8–98.0	MCR
Hb F	0.0	%	0.0–0.9	MCR
⚠ Low **Hb A2**	**1.9**	%	2.0–3.3	[1] MCR

Result Name	Value	Unit	Reference Value	Performing Site
HPLC Hb Variant, B	See Interpretation			[1] MCR
Ferritin, S	50	mcg/L	11–307	MCR

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Laboratory Notes

[1] This test has been modified from the manufacturer's instructions. Its performance characteristics were determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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