Patient ID	Patient Name		Birth Date	Gender	Age
SA00140510	VALIDATIONTESTING, THEV1		1969-09-09	F	51
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00140510	SA00140510	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Nov 2020 12:00			

Alpha-Globin Gene Analysis

Alpha-Globin Gene Analysis (ATHL)

Result Summary

MCR

NEGATIVE

Result

MCR

No deletions or duplications within the alpha globin gene cluster were identified. Neither the Hb Constant Spring nor the alphaT Saudi alterations were identified.

Interpretation

MCR

This result reduces the likelihood, but does not rule out a diagnosis of or positive carrier status for alpha thalassemia.

This assay does not detect non-deletion types of mutations, such as point mutations other than the alphaT Saudi and Hb Constant Spring. 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 and the Hb Constant Spring and alphaT Saudi point mutations within the alpha globin gene cluster(GenBank accession number NM_000517; build GRCh37 (hg19)). 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

VICTORIA WAUGH

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: 24 Nov 2020 14:55 **Reported:** 25 Nov 2020 08:29

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
SA00140510	VALIDATIONTESTING, THEV1		1969-09-09	F	51
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00140510	SA00140510	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Nov 2020 12:00			

Thalassemia Summary Interpretation

Thalassemia Summary Interpretation MCR

MOLECULAR RESULTS:

Alpha Globin Cluster Del/Dup: Negative

SUMMARY INTERPRETATION:

1) Alpha globin gene deletion/duplication testing is negative for the most common causes of alpha thalassemia (large deletional alpha globin mutations and two common non-deletional alpha thalassemia mutations, Hb Constant Spring and AlphaT Saudi). This greatly reduces the likelihood of, but does not entirely exclude, an alpha thalassemia mutation as this assay does not detect less common non-deletional alpha thalassemia mutations. Alpha globin gene sequencing is available if clinically indicated. If desired, please call the Metabolic Hematology Laboratory at 1–800–533–1710.

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 may be of benefit to assist in the interpretation of these results.

Reviewed By MCR

JENNIFER MAIN

Received: 24 Nov 2020 14:53 **Reported:** 25 Nov 2020 09:37

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
SA00140510	VALIDATIONTESTING, THEV1		1969-09-09	F	51
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00140510	SA00140510	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Nov 2020 12:00			

Thalassemia and Hemoglobinopathy Ev

Hemoglobinopathy Interpretation

[1] MCR

1) No abnormal hemoglobin variant or beta thalassemia detected by protein methods.

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

3) Alpha thalassemia deletion/duplication testing (ATHAL) is pending. See THEV0/Thalassemia Summary Interpretation for correlation of these results with protein analysis and any provided clinical phenotype.

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
Hb A	97.0	%	95.8–98.0	MCR
Hb F	0.5	%	0.0–0.9	MCR
Hb A2	2.5	%	2.0–3.3	[1] MCR

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

Received: 24 Nov 2020 14:53

Reported: 24 Nov 2020 15:14

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.

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