1-800-533-1710

MAYO CLINIC LABORATORIES

Patient ID	Patient Name		Birth Date	Sex	Age
SA01029790	MILLER, ROBERT GERON		1980-11-07	M	42
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA01029790	4400013145	FRIES,RICHARD			
Account Information		Collected			
C7032766 Tarrant County Medical Examiner Office		19 Nov 2022 09:56			

Hb Electrophoresis Summary Interp

Hb Electrophoresis Summary Interp MCR

This report is issued to summarize all testing performed under the Hemoglobin Electrophoresis Evaluation.

MOLECULAR RESULTS:

Beta Globin Sequencing: Positive

The following alteration was detected:
Gene: HBB
Legacy: Beta 6, GAG>GTG, Glu>Val
HGVS: c.20A>T, p.E7V
Genomic: g.5248232T>A
Heterozygous
Classification: Hb S mutation

SUMMARY INTERPRETATION:

The DNA sequencing results confirm heterozygous Hb S mutation. Hb S (a beta globin variant) is not typically associated with clinical or hematologic abnormalities in heterozygous individuals under normal conditions, although complications can arise (Tsaras 2009 PMID: 19393983, Naik, 2015 PMID: 26637716). No additional beta variants were detected by DNA sequencing.

Per client, this is a post-mortem blood sample. At client request, this test was performed outside of our validated test conditions; therefore, these test results should not be used for clinical diagnostic purposes. This test has only been fully validated for pre-mortem peripheral blood samples and the performance characteristics have not been established for postmortem samples. Some confirmatory reflex testing may not be possible, and some results may be outside of the normal reported range due to specimen stability considerations. If needed, testing of family members may be useful to establish the significance of reported findings.

GENETIC COUNSELING INFORMATION:

These results may have relevance for this individual's relatives or descendants. For genetic counseling purposes, Hb S is not typically associated with clinical or hematologic abnormalities in heterozygous individuals. However, Hb S poses some reproductive risk for offspring because it can cause a sickling disorder when co-inherited with Hb S, Hb C, Hb C-Harlem, Hb D-Punjab, Hb E, Hb O-Arab, Hb New York, beta thalassemia and other rare alterations in the beta globin gene cluster. A genetic consultation may be of benefit.

Reviewed By MCR

Jennifer L. Herrick, M.D.

Received: 25 Nov 2022 10:35 **Reported:** 12 Dec 2022 15:53

Hb Electrophoresis Evaluation

Hb Electrophoresis 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 Hemoglobin (Hb) Electrophoresis Summary Interpretation (Test ID HBEL0) 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.

Per client, this patient is deceased. At client request, this test was performed outside of our validated test conditions and routine quality parameters may not be met; therefore, these test results should not be used for clinical diagnostic purposes. This test has only been fully validated for premortem peripheral blood samples and the performance characteristics have not been established for postmortem samples. Some confirmatory reflex testing may not be possible and some results may be outside of

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

1-800-533-1710

MAYO CLINIC LABORATORIES

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Order Number	Client Order Number	Ordering Physician	Report Notes		
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Account Information		Collected			
C7032766 Tarrant County Medical Examiner Office		19 Nov 2022 09:56			

the normal reported range due to specimen stability considerations. If needed, testing of family members may be useful to establish the significance of reported findings.

Protein studies indicate features of marked degradation are present. There are multiple peaks, many of which are interpreted as likely degradation peaks; however, there are peaks in the Hb A and Hb S windows/zones in both HPLC and capillary electrophoresis tracings; however, they cannot be reliably quantitated. Mass spectrometry shows a peak at the mass unit expected for normal beta chains and a peak at the mass unit expected for a Hb S mutation, although other variants are not excluded. Reliable estimation of the presence of Hb F or Hb A2 is not possible. Final classification would benefit from correlation with the presence or absence of red blood cell transfusion within approximately 120 days prior to sample collection and any prior hemoglobin electrophoresis testing, if performed. In addition, per client request, DNA sequencing of the beta globin genes will be attempted.

Methodologies utilized in this interpretation include: capillary electrophoresis, HPLC, mass spectrometry

Hb Variant, A2 and F Quantitation,B

Result Name	Value	Unit	Reference Value	Performing Site
⚠ Low Hb A		%	95.8–98.0	MCR
unable to quanitate due to degradation				
Hb F		%	0.0–0.9	MCR
unable to quanitate due to degradation				
Hb A2		%	2.0–3.3	(1) MCR
unable to quanitate due to degradation				

Result Name	Value	Unit	Reference Value	Performing Site
HPLC Hb Variant, B	See Interpretation			(1) MCR

Received: 25 Nov 2022 10:35 **Reported:** 02 Dec 2022 13:09

Hb Variant by Mass Spec, B

Result Name	Value	Unit	Reference Value	Performing Site
Hb Variant by Mass Spec, B	See Interpretation			(2) MCR

Received: 25 Nov 2022 10:35 **Reported:** 01 Dec 2022 11:49

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MAYO CLINIC LABORATORIES

1-800-533-1710

WBSQR

Beta-Globin Gene Sequencing, Blood

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Account Information		Collected			
C7032766 Tarrant County Medical Examiner Office		19 Nov 2022 09:56			

Beta Globin Gene Sequencing, B

Beta Globin Gene Sequencing Result MCR

Positive. See interpretation

Interpretation (2) MCR

Beta Globin Sequencing: Positive

Gene: HBB
Legacy: Beta 6, GAG>GTG, Glu>Val
HGVS: c.20A>T, p.E7V
Genomic: g.5248232T>A
Heterozygous
Classification: Hb S mutation

See HBEL0/HB Electrophoresis Summary Interpretation for correlation of these results with protein analysis and any provided clinical phenotype. If detected, alterations classified as Benign or Likely Benign, silent or intronic variants not known or predicted to be clinically significant are not included in this report but are available upon request. Alterations are reported in Legacy and HGVS nomenclatures and by genomic location. Genetic counseling may be of benefit to assist in the interpretation of these results.

Signing Pathologist: Jennifer L. Herrick, M.D.

ADDITIONAL INFORMATION

Bi-directional sequence analysis was performed to test for the presence of a mutation in all coding regions and non-coding portions of the beta hemoglobin gene (HBB) with reported mutations. HGVS mutation nomenclature is based on human assembly GRCh37(hg19) and RefSeq accession number NM_000518.4.

Received: 05 Dec 2022 14:24 **Reported:** 12 Dec 2022 08:56

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.

(2) 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.

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