MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,MEVP A	SA00059295	47	F	SA00059295
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00059295	**Account Information**			**Report Notes**
Collected 06/23/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/25/2013 10:57				

Test	Flag	Results	Unit	Reference Value	Perform Site*
HGB Electrophoresis, Molecular					
RECEIVED: 06/24/2013 16:55 **REPORTED:** 06/25/2013 10:18					
Alpha Globin Gene Sequencing		Performed			MCR
Laboratory developed test.					
Beta Globin Gene Sequencing		Performed			MCR
Laboratory developed test.					
Alpha Globin Gene Sequence		Performed			MCR
Beta Globin Gene Sequence		Performed			MCR
Beta Globin Gene Del/Dup		Performed			MCR
Laboratory developed test.					
Manual DNA Extraction		Performed			MCR
Alpha Globin Gene Sequencing (AGGS) Beta Globin Gene Sequencing (BGGS) and Beta Globin Gene Del/Dup (BGDD) are laboratory developed tests.					
Hemoglobin F, Red Cell Distrib, B		Homocellular			MCR
RECEIVED: 06/24/2013 16:55 **REPORTED:** 06/25/2013 10:18					
-- REFERENCE VALUE --					
Reported as: Heterocellular or Homocellular					
Interpretation					MCR
Homocellular distribution of Hb F. Performed by flow cytometry.					
IEF Confirms		Performed			MCR
RECEIVED: 06/24/2013 16:55 **REPORTED:** 06/25/2013 10:18					
Hb Variant by Mass Spec, B		Performed			MCR
RECEIVED: 06/24/2013 16:55 **REPORTED:** 06/25/2013 10:18					
Laboratory developed test.					
Methemoglobinemia Evaluation					
RECEIVED: 06/24/2013 16:55 **REPORTED:** 06/25/2013 10:19					
Methemoglobinemia Interpretation					MCR
Reviewed by KENNETH SWANSON					

MET/SULF INTERPRETATION:
Elevated Methemoglobin and Sulfhemoglobin levels are due to the prescence of an M-Hemoglobin

METR INTERPRETATION:
The slight decrease in Methemoglobin Reductase is unlikely to carry clinical significance.

HB ELECTROPHORESIS INTERPRETATION:

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* Report times for Mayo performed tests are CST/CDT

TEST SITE

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An abnormal hemoglobin is present with a large amount of Hb F and no Hb A. Molecular testing was performed to clarify.

MOLECULAR RESULTS:
Alpha Gene Sequencing Results:
DNA Sequence analysis of the alpha genes did not identify a mutation associated with a hemoglobinopathy or thalassemia.

Beta Gene Sequencing Results:
DNA Sequence analysis of the Beta Genes identified Hb M-Saskatoon, a substitution at codon 63 of CAT to TAT or His to Tyr. A normal copy of the gene was not seen.

Beta Gene MLPA Results:
A Multiplex Ligand-dependent Probe Amplification (MLPA) assay of the Beta Globin Gene Complex shows a genomic deletion spanning the delta and beta gene loci.These types of deletions can be seen in either Hereditary Persistence of Fetal Hemoglobin (HPFH) or Delta-Beta Thalassemia. This case is best classified as HPFH due to the normal MCV and the degree of Hb F elevation. Clinical correlation is necessary.

INTERPRETATION:
These results are consistent with Hb M-Saskatoon with HPFH.

MOLECULAR METHODS:
Alpha Gene Sequencing Method:
Genomic DNA was extracted andSangersequencing reactions performed using primers which flank the coding and non-coding portions of the alpha-1(HBA1) and alpha-2(HBA2) genes. This method allows for detection of hemoglobinopathies and thalassemias caused by point mutationsand small insertionsor deletions.

Beta Gene Sequencing Method:
Genomic DNAwas extracted andSangersequencing reactions performed using primers which flank the coding and non-coding portions of the beta (HBB) genes. This method allows for detection of hemoglobinopathies and thalassemias caused by point mutationsand small insertionsor deletions.

Beta Gene MLPA Method:
Polymerase Chain Reaction (PCR) and Multiplex

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Ligation-dependent Probe Amplification (MLPA) were used to detect deletion-type mutations within the beta-globin gene cluster. This method uses multiple probes that hybridize throughout the beta-globin locus on chromosome 11.					
Hemoglobin A2 and F					
Hemoglobin A2	H	3.5	%	2.0-3.3	MCR
Hemoglobin F	H	41.5	%	0.0-0.9	MCR
Hemoglobin Electrophoresis, B					
Hemoglobin A	L	0.0	%	95.8-98.0	MCR
Variant		See Below	%	No abnormal variants	MCR
Result:55.0 = Hb M-Saskatoon					
Methemoglobin, B	H	3.0	%	0.0-1.5	MCR
Methemoglobin concentrations may decline 40% per day.					
Sulfhemoglobin, B	H	0.5	%	0.0-0.4	MCR
Methemoglobin Reductase, B	L	6.5	U/g Hb	6.6-13.3	MCR
Hemoglobin S, Scrn, B		Negative			MCR
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Expected result is negative					
Hemoglobin, Unstable, B	AB	Abnormal			MCR
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Expected result is normal					

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

TEST SITE