Patient ID	Patient Name		Birth Date	Gender	Age
SA00059295	SAMPLEREPORT, MEVP A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059295	SA00059295	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Jun 2013 00:00			

HGB Electrophoresis, Molecular

Result Name	Value	Unit	Reference Value	Performing Site
Alpha Globin Gene Sequencing	Performed			MCR
ADDITIONAL INFORMATION Laboratory developed test.				
Beta Globin Gene Sequencing	Performed			MCR
ADDITIONAL INFORMATION Laboratory developed test.				
Alpha Globin Gene Sequence	Performed			MCR
Beta Globin Gene Sequence	Performed			MCR
Beta Globin Gene Del/Dup	Performed			MCR
ADDITIONAL INFORMATION Laboratory developed test.				
Manual DNA Extraction	Performed			MCR

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Hemoglobin F, Red Cell Distrib, B

Result Name	Value	Unit	Reference Value	Performing Site
Hemoglobin F, Red Cell Distrib, B	Homocellular			MCR
REFERENCE VALUE Reported as: Heterocellular or Homocellular				

Interpretation MCR

Homocellular distribution of Hb F. Performed by flow cytometry.

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IEF Confirms

Result Name	Value	Unit	Reference Value	Performing Site
IEF Confirms	Performed			MCR

Received: 24 Jun 2013 16:55 **Reported:** 25 Jun 2013 10:18

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059295	SAMPLEREPORT, MEVP A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059295	SA00059295	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Jun 2013 00:00			

Hb Variant by Mass Spec, B

Result Name	Value	Unit	Reference Value	Performing Site
Hb Variant by Mass Spec, B	Performed			MCR

ADDITIONAL INFORMATION
Laboratory developed test.

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Methemoglobinemia Evaluation

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:

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 and Sanger sequencing 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 mutations and small insertions or deletions.

Beta Gene Sequencing Method:
Genomic DNA was extracted and Sanger sequencing 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 mutations and small insertions or deletions.

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

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059295	SAMPLEREPORT, MEVP A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059295	SA00059295	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Jun 2013 00:00			

Hemoglobin A2 and F

Result Name	Value	Unit	Reference Value	Performing Site
High **Hemoglobin A2**	**3.5**	%	2.0–3.3	MCR
High **Hemoglobin F**	**41.5**	%	0.0–0.9	MCR

Hemoglobin Electrophoresis, B

Result Name	Value	Unit	Reference Value	Performing Site
Low **Hemoglobin A**	**0.0**	%	95.8–98.0	MCR
Variant	55.0 = Hb M-Saskatoon	%	No abnormal variants	MCR

Result Name	Value	Unit	Reference Value	Performing Site
High **Methemoglobin, B**	**3.0**	%	0.0–1.5	MCR
Methemoglobin concentrations may decline 40% per day.				
High **Sulfhemoglobin, B**	**0.5**	%	0.0–0.4	MCR
Low **Methemoglobin Reductase, B**	**6.5**	U/g Hb	6.6–13.3	MCR

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Performing Site Legend

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MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059295	SAMPLEREPORT, MEVP A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059295	SA00059295	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Jun 2013 00:00			

Hemoglobin S, Scrn, B

Result Name	Value	Unit	Reference Value	Performing Site
Hemoglobin S, Scrn, B	Negative			MCR

REFERENCE VALUE
Expected result is negative

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Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905

Patient ID	Patient Name		Birth Date	Gender	Age
SA00059295	SAMPLEREPORT, MEVP A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059295	SA00059295	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		23 Jun 2013 00:00			

Unstable Hemoglobin, B

Hemoglobin, Unstable, B MCR

Abnormal

Abn

REFERENCE VALUE

Expected result is normal

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Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905