Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HAEVP	SA00055548	46	F	SA00055548
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00055548	**Account Information**			**Report Notes**
Collected 05/14/2013	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 05/16/2013 15:05				

Test	Flag	Results	Unit	Reference Value	Perform Site*
			REPORTED 05/16/2013 14:37		
Band 3 Fluorescence Staining, RBC	AB	Abnormal			MCR
-- REFERENCE VALUE -- Expected result is normal					
			REPORTED 05/16/2013 14:37		
Glutathione, B	H	99.9	mg/dL RBC	46.9-90.1	MCR
Hemolytic Anemia Evaluation			REPORTED 05/16/2013 14:59		
Hemolytic Anemia Interpretation					MCR

HB ELECTROPHORESIS INTERPRETATION:
Specimen was sent for molecular testing to confirm the hemoglobin variant present.

MOLECULAR RESULTS:
DNA sequence analysis of the Beta gene identifies Hb Malmo, a substitution at codon 97 of CAC to CAG, or to His to Gln.

MOLECULAR INTERPRETATION:
These results confirm Hemoglobin Malmo, a high oxygen affinity variant which causes erythrocytosis in the heterozygote.

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

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cluster. This method uses multiple probes that hybridize throughout the beta-globin locus on chromosome 11.					
RBC ENZYME INTERPRETATION: All Red Blood Cell enzyme values are normal or elevated. Elevated enzyme concentrations can be seen in patients with a younger erythrocyte population. This may be seen with reticulocytosis from any cause or in normal neonates.					
OSMOTIC FRAGILITY INTERPRETATION: Osmotic fragility testing: Increased red blood cell lysis EMA binding test (band 3 assay): Decreased fluorescence					
Interpretation: The Osmotic Fragility and Band 3 results are supportive of a diagnosis of hereditary spherocytosis. Decreased fluorescence has also been reported in other rare blood cell disorders such as hereditary pyropoikilocytosis, southeast asian ovalocytosis, congenital dyserythropoietic anemia type II and cryohydrocytosis. Therefore, correlation with the patient's clinical history, family history, and the peripheral blood smear findings is necessary.					
Hemoglobin A2 and F					
Hemoglobin A2		2.7	%	2.0-3.3	MCR
Hemoglobin F		0.3	%	0.0-0.9	MCR
Hemoglobin Electrophoresis, B					
Hemoglobin A	L	55.0	%	95.8-98.0	MCR
Variant		42.0 = Hb Malmo	%	No abnormal variants	MCR
Hemoglobin, Unstable, B		Normal			MCR
-- REFERENCE VALUE -- Expected result is normal					
Osmotic Fragility					
Osmotic Fragility, 0.50 g/dL NaCl	H	85.0	%hemol	0.0-31.1	MCR
Osmotic Fragility, 0.60 g/dL NaCl	H	89.0	%hemol	10.9-65.5	MCR
Osmotic Fragility, 0.65 g/dL NaCl	H	98.0	%hemol	0.2-39.3	MCR
Osmotic Fragility, 0.75 g/dL NaCl	H	75.0	%hemol	0.0-10.9	MCR
Sex of Control Vial		Female			MCR
G-6-PD, QN, RBC	H	18.0	U/g Hb	8.8 - 13.4	MCR
Pyruvate Kinase, RBC	H	22.3	U/g Hb	6.7 - 14.3	MCR
Glucose Phosphate Isomerase, B	H	88.3	U/g Hb	39.3 - 57.7	MCR
Hexokinase, B	H	2.0	U/g Hb	0.8 - 1.9	MCR
Morphology Review					MCR
Review of blood smear reveals a subset of spherocytes.					

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Printed 05/16/2013 15:05				

Test	Flag	Results	Unit	Reference Value	Perform Site*
HGB Electrophoresis, Molecular			REPORTED 05/16/2013 14:36		
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.					
			REPORTED 05/16/2013 14:37		
Hemoglobin F, Red Cell Distrib, B		Heterocellular			MCR
-- REFERENCE VALUE --					
Reported as: Heterocellular or Homocellular					
Interpretation					MCR
Heterocellular distribution of Hb F. Performed by flow cytometry.					
			REPORTED 05/16/2013 14:37		
IEF Confirms		Performed			MCR
			REPORTED 05/16/2013 14:38		
Hb Variant by Mass Spec, B		Performed			MCR
Laboratory developed test.					
Reflexed RBC Enzymes			REPORTED 05/16/2013 14:58		
Adenosine Deaminase, B	H	1.8	U/g Hb	0.5 - 1.7	MCR
Adenylate Kinase, B	H	325	U/g Hb	190 - 321	MCR
Phosphofructokinase, RBC	H	9.5	U/g Hb	6.1 - 9.4	MCR
Phosphoglycerate Kinase, B	H	250	U/g Hb	165 - 239	MCR
Triosephosphate Isomerase, B	H	1510	U/g Hb	930 - 1406	MCR
Pyrimidine 5' Nucleotidase, B		Normal			MCR
-- REFERENCE VALUE --					
Expected result is normal					
			REPORTED 05/16/2013 14:38		
Hemoglobin S, Scrn, B		Negative			MCR
-- REFERENCE VALUE --					
Expected result is negative					

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>> Accession SA00055548 - Continued From Previous Page <<
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* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: