

Patient ID SA00059315	Patient Name SAMPLEREPORT, THEVP A	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00059315	Client Order Number SA00059315	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 24 Jun 2013 00:00		

Alpha-Globin Gene Analysis

Result Name	Value	Unit	Reference Value	Performing Site
Alpha-Globin Gene Analysis	Performed			MCR
ADDITIONAL INFORMATION Laboratory developed test.				

Received: 25 Jun 2013 14:11

Reported: 26 Jun 2013 09:08

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

Received: 25 Jun 2013 14:11

Reported: 26 Jun 2013 09:10

Hemoglobin F, Red Cell Distrib, B

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

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905
SDL	Mayo Clinic Laboratories - Rochester Superior Drive	3050 Superior Drive NW, Rochester MN 55901



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Interpretation

MCR

Heterocellular 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: 25 Jun 2013 14:11

Reported: 26 Jun 2013 09:11

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.

Received: 25 Jun 2013 14:11

Reported: 26 Jun 2013 09:11

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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Thalassemia and Hemoglobinopathy Ev

Hemoglobinopathy Interpretation

1 MCR

Reviewed by KENNETH SWANSON

Hb E, Hb A2 and Hb F are present. Molecular testing was performed to clarify results.

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 gene identifies Hb E, a substitution at codon 26 of GAG to AAG, or Glu to Lys. 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 did not detect any large deletional genomic mutations.

AGPB Results:

Two alpha globin genes are deleted in cis, i.e., on the same chromosome.

In addition neither of the following non-deletional alpha thalassemia point mutations were found: Hb Constant Spring and the Poly A mutation AATAAA to AATAAG (also known as alphaT Saudi).

INTERPRETATION:

These results confirm Homozygous Hb E with alpha thalassemia trait.

GENETIC 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.

AGPB Method:

Dosage analysis (PCR and MLPA) was used to detect deletion-type mutations, the Hb Constant Spring, and alphaT Saudi point mutations within the alpha globin gene cluster. 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).



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Hemoglobin A2 and F

Result Name	Value	Unit	Reference Value	Performing Site
 Hemoglobin A2	5.0	%	2.0–3.3	MCR
 Hemoglobin F	30.8	%	0.0–0.9	MCR

Hemoglobin Electrophoresis, B

Result Name	Value	Unit	Reference Value	Performing Site
 Hemoglobin A	0.0	%	95.8–98.0	MCR
Variant	64.2 = Hb E	%	No abnormal variants	MCR

Result Name	Value	Unit	Reference Value	Performing Site
 Ferritin, S	6	mcg/L	11–307	SDL

Received: 25 Jun 2013 14:11

Reported: 26 Jun 2013 09:12

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Unstable Hemoglobin, B

Hemoglobin, Unstable, B

MCR



Abnormal

Abn

REFERENCE VALUE

Expected result is normal

Received: 25 Jun 2013 14:11

Reported: 26 Jun 2013 09:11

QA Environment

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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