Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,HEMP	SA00049704	19	M	SA00049704
Ordering Phys				**DOB** 10/14/1992
Client Order # SA00049704	**Account Information**			**Report Notes**
Collected 10/08/2012 12:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/30/2012 13:19				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Hereditary Erythrocytosis Mut, B			REPORTED 10/10/2012 15:09		
Molecular Interpretation					MCR

No mutations were detected in the tested genes.

This result does not exclude the presence of another alteration that may be responsible for the features present in this individual. Some individuals who have features of erythrocytosis may have a mutation that is not identified by the described testing methodology.

Genomic DNA was extracted and Sanger sequencing reactions performed to test for the presence of mutations in the Erythropoietin Receptor (EPOR) gene, exon 8 (HUGO Gene Symbol: EPOR), the Hypoxia-inducible Factor 2 Alpha (HIF2A) gene, exon 12 (HUGO Gene Symbol: EPAS1), and the Prolyl Hydroxylase Domain-2 (PHD2) gene, exons 1-5 (HUGO Gene Symbol: EGLN1).

A genetic consultation may be of benefit.

Cautions:
Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, family history, and other laboratory data, additional testing should be considered. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Rarely, individuals may have a mutation or deletion in the gene(s) tested that is not identified by the described testing methodology.

In addition, the phenotype observed in the individual tested here may be due to a variant in a gene not analyzed by this test.

EPOR Gene, Mutation Analysis, B (EPOR), HIF2A Gene, Mutation Analysis, B (HIF2A), and PHD2 Gene, Mutation

Performing Site Legend on Last Page of Report

MAYO CLINIC
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Laboratory Service Report 1-800-533-1710

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TESTINGRNV,HEMP	SA00049704	19	M	SA00049704
Ordering Phys				**DOB** 10/14/1992
Client Order # SA00049704	**Account Information**			**Report Notes**
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Printed 10/30/2012 13:19				

Test	Flag	Results	Unit	Reference Value	Perform Site*
Analysis, B (PHD2) are laboratory developed tests.					
Reviewed By		Koren Melcher			MCR
EPOR Gene, Mutation Analysis, B					
EPOR Gene Sequencing Result					MCR
Negative for EPOR gene, Exon 8 mutation by PCR and sequencing.					
HIF2A Gene, Mutation Analysis, B					
HIF2A Gene Sequencing Result					MCR
Negative for HIF2A gene, Exon 12 mutation by PCR and sequencing.					
PHD2 Gene, Mutation Analysis, B					
PHD2 Gene Sequencing Result					MCR
Negative for PHD2 gene, Exons 1-5 mutation by PCR and sequencing.					

* Performing Site:

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

* Report times for Mayo performed tests are CST/CDT