Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,HEMP	SA00049705	28	F	SA00049705
Ordering Phys				**DOB** 09/07/1984
Client Order # SA00049705	**Account Information**			**Report Notes**
Collected 10/05/2012 01:00	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 10/30/2012 13:21				

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

A heterozygous base pair deletion (-G) was detected on the PHD2 gene, exon 1, resulting in a frame shift and a truncated protein sequence. This mutation has been reported to cause erythrocytosis in a heterozygous individual. (Ref: Al-Sheikh (2008) Blood Cells Mol Dis 40, 160)

Since a mutation has been identified in the PHD2 gene in this individual, genetic testing for this specific gene in other family members is possible. Please contact the Metabolic Hematology Laboratory at 1-800-533-1710 or the on-line test catalog at www.mayomedicallaboratories.com for information about how to order the HEMP test for a family member of this individual. Please include the mutation identified in this family member with your request.

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

Performing Site Legend on Last Page of Report

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Test	Flag	Results	Unit	Reference Value	Perform Site*
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 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
Positive for PHD2 gene, Exon 1, nucleotide c.606delG, amino acid (p.Met202IlefsX71)					

* Performing Site:

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