

Patient Name SAMPLEREP, HHTM	Patient ID SA00050191	Age 46	Gender F	Order # SA00050191
Ordering Phys				DOB 06/10/1966
Client Order # SA00050191	Account Information			Report Notes
Collected 10/30/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER, MN 55901			
Printed 11/01/2012 08:39				

Test	Flag	Results	Unit	Reference Value	Perform Site*
DNA Extraction, NPL			REPORTED	10/31/2012 15:45	
Specimen		Blood			MCR
Specimen ID		1038961			MCR
Order Date		31 Oct 2012 15:29			MCR
Comment		Genomic DNA has been extracted.			MCR
Reviewed By		Sheila Marie Wilson			MCR
Release Date		31 Oct 2012 15:30			MCR

ENG and ACVRL1, Large Del/Dup			REPORTED	10/31/2012 16:42	
ENG-ACVRL1, Large Del/Dup Result					
One copy of a large genomic deletion of Exon 9 in ENG was detected (del Exon 9). A mutation in ACVRL1 was not detected.					
ENG-ACVRL1, Large Del/Dup Interp					
The presence of a hereditary hemorrhagic telangiectasia (HHT) - associated mutation in this patient places this individual at risk for developing and/or exacerbation of symptoms associated with HHT. Appropriate surveillance procedures and/or management strategies should be considered.					
<p>Since a mutation has been identified in the ENG gene in this individual, genetic testing for this specific mutation in other family members is recommended. Please contact the laboratory at 1-800-533-1710 or the on-line test catalog at mayomedicallaboratories.com for information about how to order ENG and ACVRL1, Large Del/Dup (89587). Please refer to family number X when ordering testing for family members of this individual.</p> <p>Multiplex-ligation dependent probe amplification (MLPA) was used to test for the presence of large genomic alterations in the ENG and ACVRL1 genes (GenBank numbers NM_001114753.1 and NM_000020.2, respectively). This PCR-based method utilizes probes for all 15 exons of the ENG gene, and all 10 exons of the ACVRL1 gene.</p>					
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, additional testing should be considered.					

Performing Site Legend on Last Page of Report

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* Report times for Mayo performed tests are CST/CDT

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Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. 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.

For research use only.

ENG-ACVRL1, Large Del/Dup Rev by

Jamie Bruflat

MCR

* Performing Site:

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