

Patient Name SAMPLEREP, HHTM	Patient ID SA00050190	Age 46	Gender F	Order # SA00050190
Ordering Phys				DOB 06/10/1966
Client Order # SA00050190	Account Information			Report Notes
Collected 10/30/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER, MN 55901			
Printed 11/01/2012 08:38				

Test	Flag	Results	Unit	Reference Value	Perform Site*
DNA Extraction, NPL				REPORTED 10/31/2012 15:46	
Specimen		Blood			MCR
Specimen ID		1038962			MCR
Order Date		31 Oct 2012 15:31			MCR
Comment					MCR
Genomic DNA has been extracted.					
Reviewed By		Sheila Marie Wilson			MCR
Release Date		31 Oct 2012 15:32			MCR

ENG and ACVRL1, Large Del/Dup				REPORTED 10/31/2012 16:30	
ENG-ACVRL1, Large Del/Dup Result					MCR
A mutation was not detected in ENG or ACVRL1.					
ENG-ACVRL1, Large Del/Dup Interp					MCR

This result does not rule out the diagnosis of hereditary hemorrhagic telangiectasia (HHT). Some individuals who have a diagnosis of HHT and involvement of the ENG or ACVRL1 gene may have mutations that are not identified by the described testing methodology. Furthermore, mutations in genes other than ENG or ACVRL1 may be involved in the HHT phenotype.

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.

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.

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.

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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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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Patient Name SAMPLEREP,HTM	Collection Date and Time 10/30/2012	Report Status Final
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