MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,HHTM NORM	SA00062769	33	F	SA00062769
Ordering Phys CLIENT,CLIENT				**DOB** 01/21/1980
Client Order # SA00062769	**Account Information**			**Report Notes**
Collected 09/24/2013 08:57	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 09/26/2013 13:32				

Test	Flag	Results	Unit	Reference Value	Perform Site*

ENG and ACVRL1, Large Del/Dup
RECEIVED: 09/25/2013 14:01 **REPORTED:** 09/25/2013 14:29

ENG-ACVRL1, Large Del/Dup Result MCR

The familial ENG deletion of Exon 3-8 duplication was not detected in this individual.

ENG-ACVRL1, Large Del/Dup Interp MCR

The ENG deletion of Exon 3-8 duplication was previously identified in a family member with features of a hereditary hemorrhagic telangiectasia (HHT). Since this mutation was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having HHT.

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.

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: Franklin R. Cockerill, III, M.D.

Patient Name	Collection Date and Time	Report Status
TESTINGRNV,HHTM NORM	09/24/2013 08:57	Final
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* Report times for Mayo performed tests are CST/CDT

TEST SITE