Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HHTP	0000180899	46	F	0000180899
Ordering Phys				**DOB** 06/10/1966
Client Order # 0000180899	**Account Information**			**Report Notes**
Collected 03/15/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/17/2013 10:17				

Test	Flag	Results	Unit	Reference Value	Perform Site*

ENG and ACVRL1, Full Gene Analysis
RECEIVED: 03/15/2013 00:00 **REPORTED:** 06/17/2013 09:44

ENG and ACVRL1, Full Gene Analysis

ENG and ACVRL1 Result MCR

One copy of the following mutation was detected in ENG:
Exon 3, nucleotide c.224delC, amino acid Pro75ArgfsX6. A mutation in ACVRL1 was not detected.

ENG and ACVRL1 Interpretation MCR

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.

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 Gene, Known Mutation (89391). Please refer to family number XYZ when ordering testing for family members of this individual.
Fluorescent DNA sequence analysis was used to test for the presence of mutations in all 15 exons and exon-intron boundaries of the ENG gene (GenBank number NM_001114753.1), and all 10 exons and exon-intron boundaries of the ACVRL1 gene (GenBank number NM_000020.2). In addition, multiplex-ligation dependent probe amplification (MLPA) was used to test for the presence of large genomic alterations in the ENG and ACVRL1 genes. 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.

A list of common polymorphisms identified for this patient is available from the lab upon request.

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

MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

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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.					
Laboratory developed test.					
ENG and ACVRL1 Reviewed by		Jennifer Herman			MCR
HHT Gene Sequencing		Performed			MCR
ENG and ACVRL1, Large Del/Dup		Performed			MCR
For research use only.					

* 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