Patient Name	**Patient ID**	**Age**	**Gender**	**Order #**
SAMPLEREPORT,HHTP	SA00066726	47	F	SA00066726
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00066726	**Account Information**			**Report Notes**
Collected 04/06/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 04/09/2014 08:29				

Test	Flag	Results	Unit	Reference Value	Perform Site*

ENG and ACVRL1, Full Gene Analysis
RECEIVED: 04/07/2014 12:51 **REPORTED:** 04/07/2014 14:18

ENG and ACVRL1, Full Gene Analysis

ENG and ACVRL1 Result MCR

A mutation was not detected in FBN1.

ENG and ACVRL1 Interpretation 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.
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.

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.

If the patient has had an allogeneic blood or marrow transplant or a recent (i.e. less than 6 weeks from time of sample collection) heterologous blood transfusion these results may be inaccurate due to the presence of donor DNA.
Laboratory developed test.

MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,HHTP	SA00066726	47	F	SA00066726
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00066726	**Account Information**			**Report Notes**
Collected 04/06/2014 00:00	C7028846-DLMP Rochester SDSC 2 - Client Support Rochester, MN 55901			
Printed 04/09/2014 08:29				

Test	Flag	Results	Unit	Reference Value	Perform Site*
ENG and ACVRL1 Reviewed by		CHRISTINE THOE			MCR
HHT Gene Sequencing		Performed			MCR
ENG and ACVRL1, Large Del/Dup		Performed			MCR

* Performing Site:

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

* Report times for Mayo performed tests are CST/CDT