

Patient Name SAMPLEREP, HHTP	Patient ID 0000180902	Age 46	Gender F	Order # 0000180902
Ordering Phys				DOB 06/10/1966
Client Order # 0000180902	Account Information			Report Notes
Collected 03/15/2013 00:01	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/17/2013 10:16				

Test	Flag	Results	Unit	Reference Value	Perform Site*
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ENG and ACVRL1, Full Gene Analysis
RECEIVED: 03/15/2013 00:01 **REPORTED:** 06/17/2013 09:43

ENG and ACVRL1, Full Gene Analysis

ENG and ACVRL1 Result

MCR

A mutation was not detected in ENG or ACVRL1.

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.

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.

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP, HHTP	Collection Date and Time 03/15/2013 00:01	Report Status Final
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ENG and ACVRL1 Reviewed by HHT Gene Sequencing		Jennifer Herman			MCR
ENG and ACVRL1, Large Del/Dup For research use only.		Performed			MCR
		Performed			MCR

* Performing Site:

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