MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, PAM	L3MRNG9156045	57	F	G9156045

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
09/10/10 04:48 P	09/10/10 04:48 P	09/30/10 08:50 A	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO		REF RANGE	PERFORM SITE *
DNA Extraction, NPL				
Specimen		**Blood**		**MCR**
Specimen ID		**1034622**		**MCR**
Order Date		**13 Sep 2010 08:58**		**MCR**
Comment				**MCR**
Genomic DNA has been extracted.				
Reviewed By		**Joshua A Gorman**		**MCR**
Release Date		**13 Sep 2010 08:59**		**MCR**
ENG and ACVRL1, Large Del/Dup				
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.
If sequence analysis has not already been performed for the ENG and ACVRL1 genes, it is suggested, since sequencing is expected to identify mutations in 60-80% of clinically affected individuals. For future reference, sequencing and MLPA are also available in a panel (89392, "ENG and ACVRL1, Full Gene Analysis").
Since reason for referral (RFR) was not provided, we are unable to provide a more detailed interpretation. If RFR information becomes available, please contact the laboratory at 1-800-533-1710 for a revised report.

** Perform Site Legend on last page of report*

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, PAM	Final	09/10/10 04:48 P

Specimen receipt and report times are in CST/CDT

REPRINT

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TEST REQUESTED	HI LO		REF RANGE	PERFORM SITE *
ENG-ACVRL1, Large Del/Dup Rev by		**Linnea M. Baudhuin, Ph.D.**		**MCR**

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic, Rochester MN. It has not been cleared or approved by the U.S. Food and Drug Administration.
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.
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.

*** PERFORMING SITE**

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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TESTING, PAM	Final	09/10/10 04:48 P