

1-800-533-1710

PATIENT NAME TESTING, PAM		PATIENT NUMBER L3MRNG9156046		AGE 57	SEX F	ACCESSION # G9156046
ORDERING PHYSICIAN			CLIENT ORDER #		ACCOUNT # LIAISONS	
COLLECTION 09/10/10 04:48 P	RECEIVED 09/10/10 04:48 P	REPORT PRINTED 09/30/10 08:27 A		SPECIMEN INFORMATION DATE OF BIRTH:		
DATE TIME	DATE TIME	DATE TIME				
Test Client Attn: Mayo Liaisons 200 First Street SW Rochester, MN 55905 507-284-8202						

TEST REQUESTED	HI LO	REF RANGE	PERFORM SITE *
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DNA Extraction, NPL

Specimen	Blood	MCR
Specimen ID	1034623	MCR
Order Date	13 Sep 2010 08:59	MCR
Comment	Genomic DNA has been extracted.	MCR
Reviewed By	Joshua A Gorman	MCR
Release Date	13 Sep 2010 09:00	MCR

ENG Gene, Known Mutation

ENG Result	<p>This individual was shown to be heterozygous for the following familial mutation in the ENG gene: Exon 5, nucleotide c.673C>A, amino acid p.Pro225Thr</p>	MCR
ENG Interpretation	<p>The presence of the familial mutation in this individual is consistent with a diagnosis of HHT. If features are not already present, this individual is at risk for developing symptoms associated with HHT. Appropriate surveillance procedures and/or treatment 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 online test catalog at mayomedicallaboratories.com for information about how to order ENG Gene, Known Mutation (89391) for sequencing mutations.</p>	MCR
ENG Reviewed by	Linnea M. Baudhuin, Ph.D.	MCR
<p>This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo</p>		

* Perform Site Legend on last page of report

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Clinic, Rochester MN. It has not been cleared or approved by the U.S. Food and Drug Administration. Fluorescent DNA sequence analysis was used to test for the presence of a specific alteration in the ENG gene, which was previously identified in an affected family member of this individual. 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.

ENG Known Mutation Sequencing
ENG Sequencing
Performed
MCR

* 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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