

1-800-533-1710

PATIENT NAME			PATIENT NU	MBER		AGE	SEX	ACCESSION #
TESTING, PAM			L3MRNG91	L3MRNG9156047			F	G9156047
ORDERING PHYSICIAN			CLIENT ORD	ER#	_			ACCOUNT # LIAISONS
COLLECTION	RECEIVED		REPORT PRI	NTED	SPECIMEN INFORMA	ATION		
09/10/10 04:49 P	09/10/10 0	14:49 P	09/30/10	08:27 A	DATE OF BIRTH:			
DATE TIME	DATE	TIME	DATE	TIME				
Test Client Attn: Mayo Liaisons								
200 First Street SW								
Rochester, MN 55905								
507-284-8202								
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TEST REQUESTED LO REF RANGE PERFORM SITE *

DNA Extraction, NPL

 Specimen
 Blood
 MCR

 Specimen ID
 1034624
 MCR

 Order Date
 13 Sep 2010 09:00
 MCR

 Comment
 MCR

Genomic DNA has been extracted.

 Reviewed By
 Joshua A Gorman
 MCR

 Release Date
 13 Sep 2010 09:01
 MCR

ACVRL1 Gene, Known Mutation

ACVRL1 Result MCR

This individual was shown to be heterozygous for the following familial mutation in the ACVRL1 gene:

Intron 3 (IVS3), c.313+11C>T

ACVRL1 Interpretation MCR

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 ACVRL1 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 ACVRL1 Gene, Known

Mutation (89393) for sequencing mutations.

ACVRL1 Reviewed by Linnea M. Baudhuin,

Ph.D.

This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo

* Perform Site Legend on last page of report

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, PAM	Final	09/10/10 04:49 P

MCR



1-800-533-1710

PATIENT N	IAME			PATIENT NUM	BER		AGE	SEX	ACCESSION #
TESTING, PAM			L3MRNG9156047			57	F	G9156047	
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Attn: Ma	ayo Liaisons								
200 Firs	t Street SW								
Rochest	er, MN 55905								
507-284	-8202								

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TEST REQUESTED

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REF RANGE PERFORM SITE *

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 ACVRL1 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.

ACVRL1 Known Mutation Sequencing

ACVRL1 Sequencing

Performed

MCR

* PERFORMING SITE

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

PATIENT NAME	ORDER STATUS	COLLECTION DATE AND TIME
TESTING, PAM	Final	09/10/10 04:49 P