Patient ID	Patient Name		Birth Date	Gender	Age
SA00062770	TESTINGRNV, HHTM AB		1980-01-21	F	33
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00062770	SA00062770	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		24 Sep 2013 08:58			

ENG and ACVRL1, Large Del/Dup

ENG-ACVRL1, Large Del/Dup Result

MCR

One copy of a large genomic deletion of Exon 3–8 in ENG was detected (del Exon 3–8). A mutation in ACVRL1 was not detected.

ENG-ACVRL1, Large Del/Dup Interp

MCR

The presence of a hereditary hemorrhagic telangiectasia (HHT) - associated mutation in this patient places this individual at risk for developing and/or exacerbation of symptoms associated with HHT. Appropriate surveillance procedures and/or management 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 on-line test catalog at mayomedicallaboratories.com for information about how to order ENG and ACVRL1, Large Del/Dup (89587).

ADDITIONAL INFORMATION

Multiplex-ligation dependent probe amplification (MLPA) was used to test for the presence of large genomic alterations in the ENG and ACVRL1 genes (GenBank numbers NM_001114753.1 and NM_000020.2, respectively). 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.

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.

ENG-ACVRL1, Large Del/Dup Rev by

MCR

Jamie Bruflat

Received: 25 Sep 2013 14:01 **Reported:** 25 Sep 2013 14:31

Performing Site Legend

Code	Laboratory	Address
MCR	Mayo Clinic Dept. of Lab Med and Pathology	200 First Street SW, Rochester, MN 55905