Patient ID	Patient Name		Birth Date	Gender	Age
SA00059526	SAMPLEREPORT, ACVK A		1966-06-10	F	47
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00059526	SA00059526	Client, Client			
Account Information		**Collected**			
C7028846 DLMP Rochester		27 Jun 2013 00:00			

ACVRL1 Gene, Known Mutation

ACVRL1 Gene, Known Mutation

ACVRL1 Result

MCR

One copy of the ACVRL1 Exon 10, nucleotide c.1438C>T, amino acid p.Leu480Phe (p.L480F) familial mutation was detected in this individual.

ACVRL1 Interpretation

MCR

The ACVRL1 p.L480F variant was identified at an outside laboratory in a family member of this individual. The significance of the p.L480F variant is uncertain. This variant has been previously reported in a patient who met clinical diagnostic criteria for HHT (see Olivieri et al. J Hum Genet (2007) 52:820–829). The presence of this variant in the symptomatic individual tested here suggests that the p.L480F variant is segregating with disease in this family and may provide further evidence of a pathogenic role for this variant. However, in the absence of functional studies, we are unable to definitively determine whether the p.L480F variant is a deleterious mutation or a benign polymorphism.

Given that the functional consequence of the p.L480F alteration is not definitely known, the presence of the p.L480F alteration in this individual should be interpreted in the context of phenotypic presentation, family history, and genetic testing results in other family members. Appropriate surveillance procedures and/or management strategies should be considered.

It may be useful to test for this alteration in other affected and unaffected family members to determine if this variant tracks with disease. Please contact the laboratory at 1–800–533–1710 to inquire about the ACVRL1 Known Mutation test (89393). Please reference family number XYZ when ordering known mutation testing for family members of this patient.

ADDITIONAL INFORMATION

Fluorescent DNA sequence analysis was used to test for the presence of a specific alteration in the ACVRL1 gene (GenBank number NM_000020.2), 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.

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. Laboratory developed test.

ACVRL1 Reviewed by

MCR

Jamie Bruflat

ACVRL1 Known Mutation Sequencing

MCR

Performed

Received: 03 Jul 2013 13:30 **Reported:** 04 Sep 2013 09:09

Performing Site Legend

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