



Hereditary Hemorrhagic Telangiectasia,
ACVRL1 Gene, Known Mutation

Patient ID SA00059525	Patient Name SAMPLEREPOR, ACVK N	Birth Date 1966-06-10	Gender F	Age 47
Order Number SA00059525	Client Order Number SA00059525	Ordering Physician Client, Client	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 27 Jun 2013 00:00		

ACVRL1 Gene, Known Mutation

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ACVRL1 Result

MCR

The ACVRL1 Exon 7, nucleotide c.853C>T, amino acid p.Leu285Phe (p.L285F) familial mutation was not detected in this individual.

ACVRL1 Interpretation

MCR

The ACVRL1 p.Leu285Phe mutation was previously identified in a family member with features of a hereditary hemorrhagic telangiectasia (HHT). Since this mutation was not detected in this individual, this suggests that this patient is at no greater risk than someone in the general population for having HHT.

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 13:29

Reported: 04 Sep 2013 09:06

Performing Site Legend

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