

<b>Patient Name</b> SAMPLEREP,ACVK N	<b>Patient ID</b> SA00059525	<b>Age</b> 47	<b>Gender</b> F	<b>Order #</b> SA00059525
<b>Ordering Phys</b> CLIENT,CLIENT				<b>DOB</b> 06/10/1966
<b>Client Order #</b> SA00059525	<b>Account Information</b>			<b>Report Notes</b>
<b>Collected</b> 06/27/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
<b>Printed</b> 09/16/2013 08:14				

Test	Flag	Results	Unit	Reference Value	Perform Site*
<b>ACVRL1 Gene, Known Mutation</b>					
RECEIVED: 07/03/2013 13:29 REPORTED: 09/04/2013 09:06					
ACVRL1 Gene, Known Mutation					
		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. 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.			
		Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. Laboratory developed test.			
		ACVRL1 Reviewed by	Jamie Bruflat		MCR
		ACVRL1 Known Mutation Sequencing	Performed		MCR

## \* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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<b>Patient Name</b> SAMPLEREP,ACVK N	<b>Collection Date and Time</b> 06/27/2013 00:00	<b>Report Status</b> Final
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\* Report times for Mayo performed tests are CST/CDT