Test Definition: ACVK

Hereditary Hemorrhagic Telangiectasia, ACVRL1 Gene, Known Mutation

Reporting Title: ACVRL1 Gene, Known Mutation
Performing Location: Rochester

Specimen Requirements:

This test is only applicable if a mutation has previously been identified in a family member of this individual.

Multiple cardiovascular-related gene sequencing tests can be performed on a single specimen after a single extraction. See Multiple Cardiovascular-Related Gene Sequencing Tests in Special Instructions for a list of tests that can be ordered together.

Container/Tube: Lavender top (EDTA)
Specimen Volume: 3 mL
Collection Instructions: Send specimen in original tube.
Additional Information:
1. Include physician name and phone number with the specimen.
2. Transfusions will interfere with testing for up to 4 to 6 weeks. DNA obtained from white cells may not provide useful information for patients who received a recent transfusion of blood that was not leukocyte-reduced. Wait 4 to 6 weeks until transfused cells have left the patient's circulation before drawing the patient's blood specimen for genotype testing.
Forms:
1. Hereditary Hemorrhagic Telangiectasia (HHT) Gene Testing Patient Information Sheet (Supply T650) in Special Instructions
2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.

Specimen Type	Temperature	Time
Whole Blood EDTA	Ambient (preferred)	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
31053	ACVRL1 Result	Alphanumeric		47961-8
31054	ACVRL1 Interpretation	Alphanumeric		69047-9
31055	ACVRL1 Reviewed by	Numeric		In Process
ACVKQ	ACVRL1 Known Mutation Sequencing	Alphanumeric		In Process

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Components:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
ACVKM	ACVRL1 Gene, Known Mutation	1	81479	Yes	No
ACVKQ	ACVRL1 Known Mutation Sequencing	2		Yes	No

Reference Values:

An interpretive report will be provided.