

## **NEW TEST ANNOUNCEMENT**

**NOTIFICATION DATE:** September 30, 2010 **EFFECTIVE DATE:** October 6, 2010

## HEREDITARY HEMORRHAGIC TELANGIECTASIA, ACVRL1 GENE, KNOWN MUTATION #89393

**USEFUL FOR:** Genetic testing of individuals at risk for a known activin A receptor, type II-like 1 (ACVRL1) familial mutation (associated with hereditary hemorrhagic telangiectasia). This test is only applicable if a mutation has previously been identified in a family member of this individual.

## **PROFILE INFORMATION:**

Unit Code	Reporting Name	Available Separately	Always Performed	
31053	ACVRL1 Gene, Known Mutation	No	Yes	
89441	ACVRL1 Known Mutation Sequencing	No	Yes	

**ADDITIONAL TESTS:** When this test is ordered, DNA extraction will always be performed at an additional charge.

Unit Code	Reporting Name	Available Separately	Always Performed
26968	DNA Extraction, NPL	No	Yes

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by DNA Sequence Analysis

**REFERENCE VALUES**: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: "Hereditary Hemorrhagic Telangiectasia (HHT) Gene Testing Patient Information Sheet" (Supply T650) is required for all orders.

Draw blood in a lavender-top (EDTA) tube(s), and send 3 mL of EDTA whole blood in original VACUTAINER(S).

**NOTE:** 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.
- 3. An "Informed Consent for DNA Testing" (Supply T576) is available.

## **CAUTIONS:**

- Absence of a mutation does not preclude the diagnosis of HHT unless a specific mutation has already been identified in an affected family member. Mutations in ACVRL1 and ENG occur in approximately 80% of individuals with clinically-defined HHT.
- Any error in the diagnosis or in the pedigree provided to us, including false-paternity, could lead to erroneous interpretation of results.
- Rare, undocumented polymorphisms may be present which could lead to false negative or positive results.
- Other targeted mutation analyses available: #89391, "Hereditary Hemorrhagic Telangiectasia, ENG Gene, Known Mutation" and #89587, "Hereditary Hemorrhagic Telangiectasia, ENG and ACVRL1 Large Deletion/Duplication, Molecular Analysis"

**LIST FEE:** \$700.00 for #89394 PLUS \$156.00 for #26968

**TOTAL FEE:** \$856.00

**CPT CODE**: "Hereditary Hemorrhagic Telangiectasia, ACVRL1 Gene, Known Mutation"

83898/Amplification, target, each nucleic acid sequence

83892/Enzymatic digestion

83894/Separation by gel electrophoresis

83912/Interpretation and Report

"ACVRL1 Known Mutation Sequencing": 83909/x2

"DNA Extraction, AutoPure": 83891

ANALYTIC TIME: 10 days DAY(s) SET-UP: Varies

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or Shirley Pokorski, MML Laboratory Technologist Resource Coordinator Telephone: 800-533-1710