MAYO CLINIC
Mayo Medical Laboratories

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: September 30, 2010
EFFECTIVE DATE: October 6, 2010

HEREDITARY HEMORRHAGIC TELANGIECTASIA, ENG GENE, KNOWN MUTATION #89391

USEFUL FOR: Genetic testing of individuals at risk for a known endoglin gene (ENG) familial mutation (associated with hereditary hemorrhagic telangiectasia).

NOTE: 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
31047	ENG Gene, Known Mutation	No	Yes
89439	ENG Gene, 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: #89393, "Hereditary Hemorrhagic Telangiectasia, ACVRL1 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, *ENG* Gene, Known Mutation"
83892/Enzymatic digestion
83894/Separation by gel electrophoresis
83898/Amplification, target, each nucleic acid sequence
83912/Interpretation and report

"ENG 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