MAYO CLINIC
Mayo Medical Laboratories

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

HEREDITARY HEMORRHAGIC TELANGIECTASIA, *ENG* AND *ACVRL1* LARGE DELETION/DUPLICATION, MOLECULAR ANALYSIS #89587

USEFUL FOR: Aiding in the diagnosis of hereditary hemorrhagic telangiectasia (HHT), types 1 and 2.

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: Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification

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 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:

- Related tests to detect mutations present in HHT: #89392, "Hereditary Hemorrhagic Telangiectasia, *ENG* and *ACVRL1* Full Gene Analysis" (includes both sequencing and MLPA); once a mutation has been identified in a family, targeted mutation analysis can be performed in at-risk family members: #89391 "Hereditary Hemorrhagic Telangiectasia, *ENG* Gene, Known Mutation" OR #89393, "Hereditary Hemorrhagic Telangiectasia, ACVRL1 Gene, Known Mutation".
- Absence of a mutation does not preclude the diagnosis of HHT unless a specific mutation has already been identified in an affected family member. Gene dosage mutations in *ACVRL1* and *ENG* occur in approximately 10% of individuals with clinically-defined HHT.
- This method will not detect most point mutations or small insertions or deletions, but will detect large genetic rearrangements (e.g. exon or multi-exon duplications or deletions).

- 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.

LIST FEE: $681.00 for #89587 PLUS $156.00 for #26968
TOTAL FEE: $837.00

CPT CODE:
"Hereditary Hemorrhagic Telangiectasia, ENG and ACVRL1 Large Deletion/Duplication, Molecular Analysis"

83900/Amplification, target, multiplex, first 2 nucleic acid sequences
83909/Separation and identification by high-resolution technique
83912/ Interpretation and report
83914/x29/Mutation identification by enzymatic ligation, single segment, each

"DNA Extraction, AutoPure"

83891

ANALYTIC TIME: 5 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