MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: October 5, 2012
EFFECTIVE DATE: October 10, 2012

HMBS Gene, Full Gene Analysis
Test ID: HMBSS

USEFUL FOR: Confirming a diagnosis of HMBS deficiency/acute intermittent porphyria (AIP)

METHODOLOGY: Polymerase Chain Reaction (PCR) Amplification/DNA Sequencing

REFERENCE VALUES: An interpretive report will be provided

SPECIMEN REQUIREMENTS: **Specimen must arrive within 96 hours of collection.**

Submit only 1 of the following specimens:
Preferred:

Specimen Type: Blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts
Container/Tube: T-75 or T-25 flask
Specimen Volume: 1 full T-75 or 2 full T-25 flasks
Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy
Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).
Specimen Volume: 4-mm punch
Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:

Specimen Type: Blood spot
Container/Tube: Whatman Protein Saver 903 Paper
Specimen Volume: 5 blood spots
Collection Instructions:
1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
2. Do not expose specimen to heat or direct sunlight.
3. Do not stack wet specimens.
4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

NOTE:

- Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527).
- **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 STABILITY INFORMATION:

Specimen Type	Temperature	Time
Varies	Varies	

CAUTIONS:

- A small percentage of individuals who have a diagnosis of AIP may have a mutation that is not identified by this method (eg, promoter mutations, deep intronic alterations). The absence of a mutation(s), therefore, does not eliminate the possibility of the diagnosis of AIP. For testing asymptomatic individuals it is important to first document the presence of an *HMBS* gene mutation in an affected family member.
- In some cases, DNA alterations of undetermined significance may be identified.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories at 1-800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.
- Mutations in other genes, such as *CPOX* and *PPOX* have been shown to cause other forms porphyrias. Abnormalities in these genes are not detected by this assay.

FEE: Please contact your Regional Manager for your account's fee information.

CPT CODE:
HMBS Gene, Full Gene Analysis
83891-Isolation or extraction of highly purified nucleic acid
83898 x 12-Amplification, target, each nucleic acid sequence
83909 x 24-Separation and identification by high-resolution technique
83912-Interpretation and report

Fibroblast Culture for Genetic Testing
88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

DAY(S) SET UP: Tuesday, 10 am **ANALYTIC TIME**: 10 days

NOTE: The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
Acute Porphyrias Panel (AIP, HCP and VP)	ZW222	MML502	Mount Sinai Genetic Testing Laboratory

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710