MAYO
Mayo Medical Laboratories
1-800-533-1710

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

HMBS Gene, Known Mutation
Test ID: HMBSK

USEFUL FOR:
- Diagnostic confirmation of HMBS deficiency when a familial mutation has been previously identified
- Carrier screening of at-risk individuals when a mutation in the *HMBS* gene has been identified in an affected family member

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:

- The identification of a disease-causing mutation in an affected family member is necessary before predictive testing for other family members can be offered. If a familial mutation has not been previously identified, order HMBSS/61216 *HMBS* Gene, Full Gene Analysis.
- Analysis is performed for the familial mutation provided only. This assay does not rule-out the presence of other mutations in this gene or in other genes that may be associated with AIP.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Any error in the diagnosis or in the pedigree provided to us, including false-paternity, could lead to erroneous interpretation of results.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

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

CPT CODE:
HMBS Gene, Known Mutation
83891-Isolation or extraction of highly purified nucleic acid
83898 x 1-Amplification, target, each nucleic acid sequence
83909 x 2-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