MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: August 29, 2012
EFFECTIVE DATE: August 29, 2012

Methylmalonic Aciduria and Homocystinuria, cblD Type, Full Gene Analysis

Test ID: MHDMS

USEFUL FOR:

- Confirmation of diagnosis of disorders belonging to the cblD complementation group
- Distinguishing between cblC, cblD, and cblF types when methylmalonic aciduria and homocystinuria are identified
- Distinguishing between cblA, cblB, and cblD variant 2 when methylmalonic aciduria is identified
- Distinguishing between cblD variant 1, cblE, and cblG when homocystinuria is identified
- Carrier screening in cases where there is a family history of methylmalonic aciduria or homocystinuria, but disease-causing mutations have not been identified in an affected individual

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

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/Frozen

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:

- **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.
- 2. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
varies	varies	

CAUTIONS:

- A small percentage of individuals who are carriers or have a diagnosis of methylmalonic aciduria and homocystinuria, cblD type (MMADHC) may have a mutation that is not identified by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation, therefore, does not eliminate the possibility of positive carrier status or the diagnosis of MMADHC. For carrier testing, it is important to first document the presence of a *MMADHC* 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 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.

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

CPT CODE:
Methylmalonic Aciduria and Homocystinuria, cblD Type, Full Gene Analysis
83891-Isolation or extraction of highly purified nucleic acid
83898 x 8-Amplification, target, each nucleic acid sequence
83909 x 16-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: Wednesday 10 am **ANALYTIC TIME**: 10 days

NOTE: Mayo referral test Methylmalonic Aciduria (Cbl A/Cbl B): MMAA and MMAB Full Gene Sequencing, ZW116, referral lab code MU, to Emory Genetics Laboratory will become obsolete.

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