MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: June 26, 2012
EFFECTIVE DATE: June 27, 2012

Carnitine Palmitoyltransferase II Deficiency, Known Mutation
Test ID: CPTKM

USEFUL FOR:
- Diagnostic confirmation of carnitine palmitoyltransferase II (CPT II) deficiency when familial mutations have been previously identified
- Carrier screening of at-risk individuals when a mutation in the CPT2 gene has been identified in an affected family member

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

NOTE: Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing. All prenatal specimens must be accompanied by a maternal blood specimen. Order MCC/88636 Maternal Cell Contamination, Molecular Analysis on both the prenatal and maternal specimens.

Preferred: Prenatal
Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15-mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated

Acceptable: Prenatal
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated

NOTE: Forms:

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

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 CTP2S/61120 *CPT2* Gene, Full Gene Analysis.
- Analysis is performed for the familial mutations provided only. This assay does not rule out the presence of other mutations within this gene or within other genes that may be associated with metabolic disease.
- 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.

LIST FEE: $ 635.00

- For skin biopsy specimens, the following test will be added at an additional charge:
  $261.10 for #80333 "Fibroblast Culture for Genetic Testing"
  $896.10 = Total List Fee
- For amniotic fluid specimens, the following test will be added at an additional charge:
  $587.80 for #80334 "Amniotic Fluid Culture for Genetic Testing"
  $1,222.80= Total List Fee
- For chorionic villus specimens, the following test will be added at an additional charge:
  $261.10 for #80333 "Fibroblast Culture for Genetic Testing"
  $896.10= Total List Fee

CPT CODE:
83891-Isolation or extraction of highly purified nucleic acid
83898 x 2-Amplification, target, each nucleic acid sequence
83909 x 4-Separation and identification by high-resolution technique
83912-Interpretation and report

Amniotic Fluid Culture for Genetic Testing
88235-Tissue culture for amniotic fluid (if appropriate)
88240-Cryopreservation (if appropriate)

Fibroblast Culture for Genetic Testing
88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)

DAY(S) SET UP: Monday 10 AM **ANALYTIC TIME**: 5 days

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