MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: August 3, 2012
EFFECTIVE DATE: August 7, 2012

Niemann-Pick Disease, Types A and B, Known Mutation
Test ID: NPDKM

USEFUL FOR:
- Diagnostic confirmation of Niemann-Pick disease type A or B when familial mutations have been previously identified
- Carrier screening of at-risk individuals when a mutation in the *SMPD1* gene has been identified in an affected family member

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by DNA Sequencing 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: Whole 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 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

NOTE: 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. **Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing**. Prenatal specimens can be sent Monday through Thursday and **must be received by 5:00 pm CST on Friday** in order to be processed appropriately.

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:

Specimen Type: Blood spot
Container/Tube: Whatman Protein Saver 903 Paper
Specimen Volume: 5 blood spots
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Confluent cultured cells (Prenatal)
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:

- Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions
- **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 NPDMS/61117 Niemann-Pick Disease, Types A and B, 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.

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

CPT CODE: Niemann-Pick A-B, Known Mutation
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)

Maternal Cell Contamination, B
83900 x 6-Amplification, target, multiplex, first 2 nucleic acid sequences (if appropriate)
83909 x 2-Separation and identification by high-resolution technique (if appropriate)
83912-Interpretation and report (if appropriate)

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

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