

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: August 3, 2012 **EFFECTIVE DATE:** August 7, 2012

Niemann-Pick Disease, Types A and B, Full Gene Analysis Test ID: NPDMS

USEFUL FOR:

• Confirmation of a diagnosis of Niemann-Pick disease type A or B

• Carrier screening in cases where there is a family history of Niemann-Pick disease type A or B, but disease-causing mutations have not been identified in an affected individual

METHODOLOGY: 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

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

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:

- A small percentage of individuals who are carriers or have a diagnosis of Niemann-Pick disease type A or B 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 Niemann-Pick disease type A or B. For carrier testing, it is important to first document the presence of an *SMPD1* gene mutation in an affected family member; however, when no mutations are detected by NPD/85322 Niemann-Pick Disease, Types A and B, Mutation Analysis, this test (NPDMS) should be ordered.
- 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: Niemann-Pick A-B Full Gene Analysis 83891-Isolation or extraction of highly purified nucleic acid 83898 x 11-Amplification, target, each nucleic acid sequence 83909 x 22-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: Thursday 10 AM

ANALYTIC TIME: 10 days

NOTE: Mayo referral tests Ashkenazi Jewish SMPD1 only from Ambry and SMPD1 Gene Analysis in Niemann-Pick Disease, Types A and B from GeneDX will become obsolete on August 7, 2012.

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