



MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: June 12, 2013

EFFECTIVE DATE: June 12, 2013

Mucopolysaccharidosis IIIB, Full Gene Analysis Test ID: MP3BS

USEFUL FOR:

- Identifying mutations within the NAGLU gene
- Confirmation of a diagnosis of mucopolysaccharidosis type IIIB
- Carrier testing when there is a family history of mucopolysaccharidosis type IIIB, but disease-causing mutations have not been previously identified

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: Whole 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:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: Ahlstrom 226 filter paper or Supplemental Newborn Screening Card (Supply T493)

Specimen Volume: 2 to 5 blood spots on collection card

Collection Instructions:

1. An alternative blood collection option for a patient >1 year of age is finger stick.
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry

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 mucopolysaccharidosis type IIIB (MPS-IIIB) 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 MPS-IIIB. The preferred approach to carrier testing is to first document the presence of an NAGLU 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.

CPT CODE:

- 81479-Unlisted molecular pathology procedure
- 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 a.m.

ANALYTIC TIME: 14 days

NOTE: The following referral test code(s) will become obsolete.

Test Name	Test ID	Referral Lab Code	Referral Lab
Mucopolysaccharidosis Type IIIB Full Gene Sequencing	ZW116	BB	Emory Genetics Laboratory

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