



**MAYO**  
Mayo Medical Laboratories  
1-800-533-1710

**NEW TEST ANNOUNCEMENT**  
**NOTIFICATION DATE:** November 14, 2013  
**EFFECTIVE DATE:** November 19, 2013

## **Mucopolysaccharidosis IIIA, Known Mutation**

Test ID: MP3AK

### **USEFUL FOR:**

- Carrier testing of individuals with a family history of mucopolysaccharidosis type IIIA
- Diagnostic confirmation of mucopolysaccharidosis type IIIA when familial mutations have 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 flask or 2 full T-25 flasks

**Specimen Stability Information:** Ambient (preferred)/Refrigerated <24 hours

**NOTE: 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 p.m. CST on Friday** in order to be processed appropriately. All prenatal specimens must be accompanied by a maternal blood specimen. Order MCC/88636 Maternal Cell Contamination, Molecular Analysis on the maternal specimen.

**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

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**ACCEPTABLE:**

**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

**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

**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 (Whatman Protein Saver 903 Paper; Ahlstrom 226 filter paper; or Supplemental Newborn Screening Card, Supply T493)  
**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:**

- 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 MP3AS/61820 Mucopolysaccharidosis IIIA, Full Gene Analysis.
- Analysis is performed only for the provided familial mutations. 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.
- We strongly recommend that patients undergoing predictive testing receive genetic counseling both prior to testing and after results are available.
- 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.

**CPT CODE:**

81479-Unlisted molecular pathology procedure code

## Additional tests:

Fibroblast Culture for Genetic Testing

88233-Tissue culture, skin, or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

Amniotic Fluid Culture for Genetic Testing

88235-Tissue culture for amniotic fluid (if appropriate)

88240-Cryopreservation (if appropriate)

Maternal Cell Contamination, Molecular Analysis

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

**DAY(S) SET UP:** Thursday, 10 a.m.

**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