

NEW TEST ANNOUNCEMENT

NOTIFICATION DATE: March 6, 2013 **EFFECTIVE DATE:** March 12, 2013

Hurler Syndrome, Known Mutation

Test ID: HURLK

USEFUL FOR:

- Carrier testing of individuals with a family history of mucopolysaccharidosis type I (MPS-I)
- Diagnostic confirmation of MPS-I when familial mutations have been previously identified

METHODOLOGY: Polymerase Chain Reaction (PCR) Amplification/DNA

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:

Invert several times to mix blood.
 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

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

Specimen Volume: 20 mg

Specimen Stability Information: Refrigerated

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: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

- 1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
- 2. Do not expose specimen to heat or direct sunlight.
- 3. Do not stack wet specimens.
- 4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Note:

- 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) 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 HURLS/61481 Hurler Syndrome, 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

Maternal Cell Contamination, Molecular Analysis

81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pretransplant recipient and donor germline testing, posttransplant 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)

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: 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