MAYO
Mayo Medical Laboratories
1-800-533-1710

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: March 6, 2013
EFFECTIVE DATE: March 12, 2013

Hurler Syndrome, Full Gene Analysis
Test ID: HURLS

USEFUL FOR:
- Identifying mutations within the *IDUA* gene
- Confirmation of a diagnosis of mucopolysaccharidosis type I (MPS-I)
- Carrier testing when there is a family history of MPS- I, but disease-causing mutations have not been previously identified

METHODOLOGY: Polymerase Chain Reaction (PCR) Followed by DNA Sequence 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
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

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
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:

- A small percentage of individuals who are carriers or have a diagnosis of mucopolysaccharidosis type I (MPS-I) 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-I. The preferred approach to carrier testing is to first document the presence of an *IDUA* 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

DAY(S) SET UP: Thursday, 10 AM **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 I – Full Gene Sequencing	ZW116	BO	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