
Reporting Title: Hurler Syndrome, Full Gene Analysis

Performing Location: Rochester

Ordering Guidance:

First-tier testing for mucopolysaccharidosis type I is available. Order either IDUAW / Alpha-L-Iduronidase, Leukocytes or PLSD / Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot. Be aware that these tests are not reliable for carrier testing.

For diagnostic testing or monitoring ongoing therapy, order MPSBS / Mucopolysaccharidosis, Blood Spot.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.

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 whole blood specimen in original tube. **Do not aliquot.**

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

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

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.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Acceptable:

Specimen Type: Blood spot

Supplies: Card - Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) filter paper, or blood spot collection card

Specimen Volume: 2 to 5 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 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

Additional Information:

1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.
2. For collection instructions, see [Blood Spot Collection Instructions](#)
3. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777)
4. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800)

Forms:

1. [New York Clients-Informed consent is required](#). Document on the request form or electronic order that a copy is on file. The following documents are available in Special Instructions:
 - [Informed Consent for Genetic Testing](#) (T576)
 - [Informed Consent for Genetic Testing-Spanish](#) (T826)
2. [Molecular Genetics: Biochemical Disorders Patient Information](#) (T527) in Special Instructions

| Specimen Type | Temperature | Time | Special Container |
|---------------|-------------|------|-------------------|
| Varies | Varies | | |

Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|-----------|------------------------|--------------|------|---------|
| 53950 | Result Summary | Alphanumeric | | 50397-9 |
| 53951 | Result | Alphanumeric | | 82939-0 |
| 53952 | Interpretation | Alphanumeric | | 69047-9 |
| 53953 | Additional Information | Alphanumeric | | 48767-8 |
| 53954 | Specimen | Alphanumeric | | 31208-2 |
| 53955 | Source | Alphanumeric | | 31208-2 |
| 53956 | Released By | Alphanumeric | | 18771-6 |

LOINC® and CPT codes are provided by the performing laboratory.

Supplemental Report:
No

CPT Code Information:
81406 IDUA (iduronidase, alpha-L-) (eg, mucopolysaccharidosis type I), full gene sequence
88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)

Reflex Tests:

| Test ID | Reporting Name | CPT Units | CPT Code | Always Performed | Available Separately |
|---------|-------------------------------------|-----------|----------|------------------|----------------------|
| CULFB | Fibroblast Culture for Genetic Test | 1 | 88233 | No | Yes |

Result Codes for Reflex Tests:

| Test ID | Result ID | Reporting Name | Type | Unit | LOINC® |
|---------|-----------|------------------------|--------------|------|---------|
| CULFB | 52327 | Result Summary | Alphanumeric | | 50397-9 |
| CULFB | 52329 | Interpretation | Alphanumeric | | 69965-2 |
| CULFB | 52328 | Result | Alphanumeric | | 82939-0 |
| CULFB | CG770 | Reason for Referral | Alphanumeric | | 42349-1 |
| CULFB | CG899 | Specimen | Alphanumeric | | 31208-2 |
| CULFB | 52331 | Source | Alphanumeric | | 31208-2 |
| CULFB | 52332 | Method | Alphanumeric | | 85069-3 |
| CULFB | 54625 | Additional Information | Alphanumeric | | 48767-8 |
| CULFB | 52333 | Released By | Alphanumeric | | 18771-6 |

Reference Values:
An interpretive report will be provided.