
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 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

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

Acceptable:

Specimen Type: Blood spot

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

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: 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 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

Additional Information:

1. For collection instructions, see [Blood Spot Collection Instructions](#) in Special Instructions.
2. For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777) in Special Instructions.
3. For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800) in Special Instructions.

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 (preferred)	0 hours	

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
FIBR	Fibroblast Culture	1	88233	No	Yes
CRYOB	Cryopreserve for Biochem Studies	1	88240	No	No

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC
CRYOB	88832	Cryopreserve for Biochem Studies	Alphanumeric		No LOINC Needed
FIBR	8482	Fibroblast Culture	Alphanumeric		96300-9

Reference Values:

An interpretive report will be provided.