Test Definition: IDUAW

Alpha-L-Iduronidase, Leukocytes

Overview

Useful For

Diagnosis of mucopolysaccharidosis I, Hurler, Scheie, and Hurler-Scheie syndromes in leukocytes

This test is **not useful** for determining carrier status.

Genetics Test Information

This test provides diagnostic testing for patients with clinical signs and symptoms suspicious for mucopolysaccharidosis type I (MPS I).

Enzyme testing is included in the diagnostic workup for infants following a positive newborn screen result for MPS I.

Testing Algorithm

Additional information is available:

- Lysosomal Disorders Diagnostic Algorithm, Part 1

-Newborn Screen Follow-up for Mucopolysaccharidosis Type I Decreased Alpha-L-Iduronidase Activity

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Newborn Screen Follow-up for Mucopolysaccharidosis Type I Decreased Alpha-L-Iduronidase Activity
- Informed Consent for Genetic Testing (Spanish)
- Lysosomal Disorders Diagnostic Algorithm, Part 1

Method Name

Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD

Ordering Guidance

This test is preferred for diagnostic testing. For carrier detection, order IDUA / Mucopolysaccharidosis Type I, *IDUA* Gene Sequencing with Deletion/Duplication, Varies.

Shipping Instructions

For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerated within 6 days of collection to

be stabilized. Pre-analytical processing is performed Monday through Friday and Sunday. This test may be canceled if specimens are outside of stability when processing occurs. Collect and package specimens for arrival on days when processing is performed.

Specimen Required

Container/Tube:
Preferred: Yellow top (ACD solution B)
Acceptable: Yellow top (ACD solution A) or lavender top (EDTA)
Specimen Volume: 6 mL
Collection Instructions: Send whole blood specimen in original tube. **Do not aliquot**.

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:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Biochemical Genetics Patient Information (T602)
3. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Minimum Volume

4 mL

Reject Due To

Gross hemolysis	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole Blood ACD	Refrigerated (preferred)	6 days	
	Ambient	6 days	

Clinical & Interpretive

Clinical Information

The mucopolysaccharidoses (MPS) are a group of lysosomal disorders caused by the deficiency of any of the enzymes involved in the stepwise degradation of dermatan sulfate, heparan sulfate, keratan sulfate, or chondroitin sulfate, also known as glycosaminoglycans (GAG) or mucopolysaccharides. Accumulation of GAG in lysosomes interferes with normal functioning of cells, tissues, and organs. There are 11 known disorders that involve the accumulation of GAG. MPS disorders involve multiple organ systems and are characterized by coarse facial features, cardiac abnormalities, organomegaly, intellectual disabilities, short stature, and skeletal abnormalities.

Mucopolysaccharidosis I (MPS I) is an autosomal recessive disorder caused by reduced or absent activity of the enzyme alpha-L-iduronidase due to variants in the *IDUA* gene. Deficiency of alpha-L-iduronidase can result in a wide range of phenotypes categorized into 3 syndromes: Hurler syndrome (MPS IH), Scheie syndrome (MPS IS), and Hurler-Scheie syndrome (MPS IH/S). Because these syndromes cannot be distinguished biochemically, they are also referred to as MPS I and attenuated MPS I.

Clinical features and severity of symptoms of MPS I are variable, ranging from severe disease to an attenuated form that generally presents at a later onset with a milder clinical presentation. In general, symptoms may include coarse facies, progressive dysostosis multiplex, hepatosplenomegaly, corneal clouding, hearing loss, intellectual disabilities or learning difficulties, and cardiac valvular disease. The incidence of MPS I is approximately 1 in 100,000 live births. Treatment options include hematopoietic stem cell transplantation and enzyme replacement therapy.

Individuals with MPS I typically demonstrate elevated levels of the GAG dermatan sulfate and heparan sulfate (see MPSQU / Mucopolysaccharides Quantitative, Random, Urine; MPSWB / Mucopolysaccharides, Blood). Reduced or absent activity of alpha L-iduronidase can confirm a diagnosis of MPS I; however, enzymatic testing is not reliable for carrier detection. Molecular sequence analysis of the *IDUA* gene allows for detection of a disease-causing variant in affected individuals and subsequent carrier detection in relatives (see IDUA / Mucopolysaccharidosis Type I, *IDUA* Gene Sequencing with Deletion/Duplication, Varies). To date, a clear genotype-phenotype correlation has not been established.

Reference Values

> or =2.06 nmol/hour/mg protein
An interpretive report will be provided.

Interpretation

Results below 2.06 nmol/hour/mg protein in properly submitted specimens are consistent with alpha-L-iduronidase deficiency (mucopolysaccharidosis I). Further differentiation between Hurler, Scheie, and Hurler-Scheie syndromes is dependent upon the clinical findings.

Normal results (> or =2.06 nmol/hour/mg protein) are not consistent with alpha-L-iduronidase deficiency.

Cautions

The presence of a pseudodeficiency allele may cause reduced activity of alpha-L-iduronidase in the artificial substrate used in this assay. This can result in values below the normal reference range but will typically be greater than levels found in individuals with mucopolysaccharidosis I (MPS I).

This test does not differentiate between Hurler and Scheie syndromes.

Enzyme levels may be normal in individuals receiving enzyme replacement therapy or who have undergone hematopoietic stem cell transplant.

Clinical Reference

1. Newborn Screening ACT Sheet [alpha-L-iduronidase deficiency with or without glycosaminoglycan (GAG) accumulation] Mucopolysaccharidosis Type I (MPS I). American College of Medical Genetics and Genomics; 2023. Updated November 2023. Accessed July 22, 2025. Available at www.acmg.net/PDFLibrary/MPSI-ACT-Sheet.pdf
2. Clark LA, Atherton AM, Burton BK, et al. Mucopolysaccharidosis type I newborn screening: Best practices for diagnosis

and management. J Pediatr. 2017;182:363-370
3. Martins AM, Dualibi AP, Norato D, et al. Guidelines for the management of mucopolysaccharidosis type I. J Pediatr. 2009:155(4 Suppl):S32-S46
4. Enns GM, Steiner RD, Cowan TM: Lysosomal disorders: mucopolysaccharidoses. In: Sarafoglou K, Hoffmann GF, Roth KS, eds. Pediatric Endocrinology and Inborn Errors of Metabolism. McGraw-Hill, Medical Publishing Division; 2009:721-730
5. Clarke LA: Mucopolysaccharidosis type I. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2002. Updated April 11, 2024. Accessed July 22, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK1162/
6. Elliott S, Buroker N, Cournoyer JJ, et al. Pilot study of newborn screening for six lysosomal storage diseases using tandem mass spectrometry. Mol Genet Metab. 2016;118(4):304-309

Performance

Method Description
The specimens are incubated with a mix of substrate and internal standard for acid sphingomyelinase, beta-glucocerebrosidase, acid alpha-glucosidase, alpha-galactosidase, galactocerebrosidase, and alpha-L-iduronidase. The sample is then purified by liquid-liquid extraction. The extract is evaporated and reconstituted before analysis by tandem mass spectrometry.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Preanalytical processing: Monday through Friday, Sunday
Testing performed: Monday, Thursday

Report Available
2 to 5 days

Specimen Retention Time
White blood cell homogenate: 1 month

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact Customer Service.

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Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information

82657

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
IDUAW	Alpha-L-Iduronidase, Leukocytes	24057-2

Result ID	Test Result Name	Result LOINC® Value
606276	Alpha-L-Iduronidase, Leukocytes	24057-2
606277	Interpretation	59462-2
606278	Reviewed By	18771-6