Test Definition: GUSBW

Beta-Glucuronidase, Leukocytes

Overview

Useful For

Supporting the biochemical diagnosis of mucopolysaccharidosis type VII (MPS VII, Sly syndrome) in whole blood

This test is **not useful for** carrier detection for MPS VII.

Genetics Test Information

This test provides diagnostic testing for individuals with clinical signs and symptoms suspicious for mucopolysaccharidosis type VII (MPS VII, Sly syndrome). If an enzyme deficiency is detected by this test, additional biochemical or molecular testing is required to confirm a diagnosis.

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Informed Consent for Genetic Testing (Spanish)

Method Name

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

NY State Available

Yes

Specimen

Specimen Type

Whole Blood ACD

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.

Necessary Information

1. Patient's age is required.

2. Reason for testing is required.

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 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

5 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

Mucopolysaccharidosis VII (MPS VII, Sly syndrome) is an autosomal recessive lysosomal storage disorder caused by the deficiency of beta-glucuronidase. Sly syndrome is one of the least common mucopolysaccharidoses with an incidence of 1 in 250,000 live births. Clinical features and severity of symptoms of MPS VII are widely variable ranging from severe lethal hydrops fetalis to more mild forms which generally present with later onset and a milder clinical presentation. In general, symptoms may include skeletal anomalies, coarse facies, hepatomegaly, neurological issues, and intellectual disability. Treatment utilizing enzyme replacement therapy is available for MPS VII.

A diagnostic workup for MPS includes glycosaminoglycan (GAG) determination in urine (MPSQU / Mucopolysaccharides Quantitative, Random, Urine) or blood (MPSBS / Mucopolysaccharidosis, Blood Spot, or MPSER / Mucopolysaccharides Quantitative, Serum) and molecular genetic analysis of the relevant gene. For MPS VII, molecular analysis of the *GUSB* gene (CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify Gene List ID: IEMCP-L613TF) allows for detection of disease-causing variants in affected patients and subsequent carrier detection in relatives.

Reference Values

>3.50 nmol/h/mg protein

An interpretive report will be provided.

Interpretation

Abnormal results are not sufficient to establish a diagnosis of a particular disease. To verify a preliminary diagnosis based on this assay, additional biochemical or molecular genetic analyses are required.

When abnormal results are detected, a detailed interpretation is given, including an overview of the results and of their significance, a correlation to available clinical information, elements of differential diagnosis, recommendations for additional biochemical testing, and in vitro, confirmatory studies (enzyme assay, molecular analysis), and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

Individuals with pseudodeficiency alleles can show reduced enzyme activity.

Carrier status (heterozygosity) for these conditions cannot be reliably detected.

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

Clinical Reference

1. Neufeld EF, Muenzer J. The mucopolysaccharidoses. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw-Hill; September 10, 2025. https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225544161
2. Sun A, Wang R. Mucopolysaccharidosis Type VII. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, eds. GeneReviews [Internet]. University of Washington, Seattle; 2024; 1993-2025 Accessed September 11, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK598990/

Performance

Method Description

Leukocytes are incubated with four cocktail mixes in 96-well plates: 1) substrate and internal standard (IS) for iduronate 2-sulfatase, heparan N-sulfatase, alpha-N-acetylglucosaminidase, N-acetylgalactosamine-sulfate, beta-galactosidase, arylsulfatase B, beta-glucuronidase, and tripeptidyl peptidase 1; 2) substrate and IS for acetyl-CoA:alpha-glucosaminide N-acetyltransferase; 3) substrate and IS for N-acetylglucosamine-6-sulfatase; and 4) substrate and IS for palmitoyl-protein thioesterase 1. Following overnight incubation, the plates are combined and purified by liquid-liquid extraction. The extracts are evaporated, reconstituted with mobile phase, and analyzed by tandem mass spectrometry.(Unpublished Mayo method)

PDF Report

No

Day(s) Performed

Preanalytical processing: Monday through Friday, Sunday
Testing performed: Tuesday

Report Available
3 to 9 days

Specimen Retention Time
WBC 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.

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
GUSBW	Beta-Glucuronidase, WBC	24065-5

Result ID	Test Result Name	Result LOINC® Value
BG763	Reason for Referral	42349-1
618289	Beta-glucuronidase	24065-5
618452	Interpretation	59462-2
618451	Reviewed By	18771-6