# Test Definition: GUSBB

Beta-Glucuronidase, Blood Spot

## Overview

### Useful For

Supporting the biochemical diagnosis of mucopolysaccharidosis VII (MPS VII, Sly syndrome)

This test is **not useful** for determining carrier status 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
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Informed Consent for Genetic Testing (Spanish)
- Blood Spot Collection Instructions

### Method Name

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

### NY State Available

Yes

## Specimen

### Specimen Type

Whole blood

### Necessary Information

**1. Patient's age is required.**

**2. Reason for testing is required.**

### Specimen Required

**Submit only 1 of the following specimen types:**

**Preferred:**

**Specimen Type:** Blood spot

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

**Container/Tube:**

**Preferred:** Blood Spot Collection Card
**Acceptable:** Whatman Protein Saver 903 Paper, PerkinElmer 226 filter paper, Munktell filter paper, or blood collected in tubes containing EDTA and dried on filter paper
**Specimen Volume:** 2 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 a Dried Blood Spot Sample.
2. At least 2 spots should be complete (ie, unpunched).
3. Let blood dry on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. Keep specimen dry.
**Specimen Stability Information:** Refrigerated (preferred) 60 days/Ambient 7 days/Frozen 60 days
**Additional Information:**
1. For collection instructions, see Blood Spot Collection Instructions
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777)
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800)

**Acceptable:**
**Specimen Type:** Whole Blood
**Container/Tube:**
**Preferred:** Lavender top (EDTA)
**Acceptable:** Yellow top (ACD)
**Specimen Volume:** 2 mL
**Collection Instructions:** Send whole blood specimen in original tube**. Do not aliquot.**
**Specimen Stability Information:** Refrigerate (preferred) 7 days/Ambient 48 hours.

## 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

Blood spot: 1; Whole blood: 0.5 mL

## Reject Due To

| | |
|---|---|
| Blood spot specimen that shows serum rings or has multiple layers | Reject |

| | |
|---|---|
| Insufficient specimen | Reject |
| Unapproved filter papers | Reject |

### Specimen Stability Information

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

## 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 1 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

>2.60 nmol/mL/h

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 genetic 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; Accessed September 11, 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. GeneReview [Internet].University of Washington, Seattle; 1993-2025. Accessed September 11, 2025. Available at www.ncbi.nlm.nih.gov/books/NBK598990/

## Performance

### Method Description

One dried blood spot (DBS) sample is incubated with a mix of 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. A second DBS sample is incubated with a mix of substrate and IS for acetyl-CoA:alpha-glucosaminide N-acetyltransferase; and a third DBS sample with a mix of substrate and IS for palmitoyl-protein thioesterase 1. Following overnight incubation, the samples are combined, extracted by liquid-liquid extraction, and analyzed by tandem mass spectrometry.(Unpublished Mayo method)

### PDF Report

No

### Day(s) Performed

Thursday

### Report Available

3 to 9 days

### Specimen Retention Time

1 year

### 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 |
|---|---|---|
| GUSBB | Beta-Glucuronidase, BS | 79457-8 |

| Result ID | Test Result Name | Result LOINC® Value |
|---|---|---|
| BG747 | Reason for Referral | 42349-1 |
| 618288 | Beta-glucuronidase | 79457-8 |
| 618428 | Interpretation | 59462-2 |
| 618427 | Reviewed By | 18771-6 |