



Beta-Glucuronidase, Leukocytes

Test ID: GUSBW

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.

Genetics 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.

Methods:

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

Reference Values:

>3.50 nmol/hour/mg protein

An interpretive report will be provided.

Specimen Requirements:

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.
Minimum Volume:	5 mL

Specimen Stability Information:

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

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.

CPT Code:

82657

Day(s) Performed:

Preanalytical processing: Monday through Saturday

Testing performed: Tuesday

Report Available:

8 to 15 days

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.