

Overview

Useful For

Supporting the biochemical diagnosis of mucopolysaccharidoses types IIIA, IIIB, IIIC, IIID

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

Genetics Test Information

This panel provides diagnostic testing for individuals with clinical signs and symptoms suspicious for mucopolysaccharidosis type IIIA, IIIB, IIIC or IIID. 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. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

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 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

5 mL

Reject Due To

Gross hemolysis	Reject
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Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

Mucopolysaccharidosis III (MPS III; Sanfilippo syndrome) is caused by reduced or absent activity of 1 of 4 enzymes involved in heparan sulfate degradation. Patients with MPS III uniformly excrete heparan sulfate resulting in similar clinical phenotypes and are further classified as type A, B, C, or D based upon the specific enzyme deficiency. MPS III is characterized by severe central nervous system (CNS) degeneration but only mild physical disease. Such disproportionate involvement of the CNS is unique among the MPS. Onset of clinical features, most commonly behavioral problems and delayed development, usually occurs between 2 and 6 years in a child who previously appeared normal. Severe neurologic degeneration occurs in most patients by 6 to 10 years accompanied by a rapid deterioration of social and adaptive skills with death generally occurring by their 20s. The occurrence of MPS III varies by subtype with types A and B being the most common and types C and D being very rare. This assay detects all MPSIII types (MPS IIIA, IIIB, IIIC, and IIID).

A diagnostic workup for MPS typically also includes glycosaminoglycan 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(s). For MPS III, a molecular panel is available that includes *SGSH*, *NAGLU*, *GNS*, *HGSNAT* (CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify Gene List ID: IEMCP-7YM613).

Reference Values

Heparan-N-sulfatase:

>0.13 nmol/h/mg protein

N-acetyl-alpha-D-glucosaminidase:

>0.09 nmol/h/mg protein

Heparan-alpha-glucosaminide N-acetyltransferase:

>0.24 nmol/h/mg protein

N-acetylglucosamine-6-sulfatase:

>0.03 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; Accessed September 10, 2025. <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225544161>
2. Hopwood JJ, Ballabio A. Multiple sulfatase deficiency and the nature of the sulfatase family. 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 10, 2025. <https://ommbid.mhmedical.com/content.aspx?bookid=2709§ionid=225546905>

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 Saturday

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
MPS3W	MPS III (Four) Panel, WBC	104072-4

Result ID	Test Result Name	Result LOINC® Value
BG767	Reason for Referral	42349-1
618456	Heparan-N-sulfatase	24086-1
618457	N-acetyl-alpha-D-glucosaminidase	24092-9
618458	Heparan-alpha-glucosaminide N-acetyltransferase	24044-0
618459	N-acetylglucosamine-6-sulfatase	24098-6
618460	Interpretation	59462-2
618455	Reviewed By	18771-6