MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: September 18, 2023
Effective Date: October 17, 2023

Mucopolysaccharidoses, Nine-Enzyme Panel, Leukocytes

Test ID: MP9W

Useful for:
Supporting the biochemical diagnosis of mucopolysaccharidoses types II, IIIA, IIIB, IIIC, IIID, IVA, IVB, VI, and VII, and of multiple sulfatase deficiency.

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

Ordering Guidance:
To evaluate newborn patients in follow-up to an abnormal newborn screen for MPS I, the recommended tests are IDUAW / Alpha-L-Iduronidase, Leukocytes and MPSBS / Mucopolysaccharidosis, Blood Spot, MPSWB / Mucopolysaccharidosis, Blood), MPSER / Mucopolysaccharides Quantitative, Serum or MPSQU / Mucopolysaccharides Quantitative, Random, Urine.

To evaluate newborn patients in follow-up to an abnormal newborn screen for MPS II, the recommended tests are I2SB / Iduronate-2-Sulfatase, Blood Spot or I2SWB / Iduronate-2-Sulfatase, Leukocytes and MPSBS / Mucopolysaccharidosis, Blood Spot, MPSWB / Mucopolysaccharidosis, Blood, MPSER / Mucopolysaccharides Quantitative, Serum or MPSQU / Mucopolysaccharides Quantitative, Random, Urine.

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

Reference Values:
Heparan-N-sulfatase: >0.13 nmol/hour/mg protein

N-acetyl-alpha-D-glucosaminidase: >0.09 nmol/hour/mg protein

Heparan-alpha-glucosaminide N-acetyltransferase: >0.24 nmol/hour/mg protein

N-acetylglucosamine-6-sulfatase: >0.03 nmol/hour/mg protein

N-acetylgalactosamine-6-sulfatase: >1.60 nmol/hour/mg protein

Beta-galactosidase: >0.28 nmol/hour/mg protein

Arylsulfatase B: >0.34 nmol/hour/mg protein

Beta-glucuronidase: >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 whole blood 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:

Beta-galactosidase is reduced in patients with galactosialidosis. Those patients will also demonstrate deficient activity of neuraminidase which is not evaluated on this panel. If there is clinical suspicion of galactosialidosis, order test OLIGU / Oligosaccharide Screen, Random, Urine.

Mucolipidosis II (MLII, I-cell disease) may not be detectable by this assay. If there is clinical suspicion of MLII, please order test LSDS / Lysosomal Storage Disorders Screen, Random, Urine, NAGS / Hexosaminidase A and Total Hexosaminidase, Serum, and/or molecular genetic analysis of the GNPTAB gene, test CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies, Gene List ID: IEMCP-L5VNMC)

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.