MAYO CLINIC LABORATORIES

NEW TEST

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

Multiple Sulfatase Deficiency, Leukocytes

Test ID: MSDW

Useful for:

Supporting the biochemical diagnosis of multiple sulfatase deficiency in whole blood specimens

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

Genetics Information:

This test is a screening panel for individuals with clinical signs and symptoms suspicious for multiple sulfatase deficiency. If an enzyme deficiency is detected by this screening test, additional biochemical or molecular testing is required to confirm a diagnosis.

Methods:

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

Reference Values:

Iduronate-2-sulfatase: >2.20 nmol/hour/mg protein

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

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

N-acetylgalactosamine-6-sulfatase: >1.60 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	Refrigerated (preferred)	6 days
	Ambient	

Cautions:

Individuals with pseudodeficiency alleles can show reduced enzyme activity.

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

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.