MAYO CLINIC LABORATORIES

# Test Definition: ARSBW

Arylsulfatase B, Leukocytes

## Overview

### Useful For

Supporting the biochemical diagnosis of mucopolysaccharidosis type VI (MPS VI, Maroteaux-Lamy syndrome) in whole blood specimens

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

### Genetics Test Information

This test provides diagnostic testing for individuals with clinical signs and symptoms suspicious for mucopolysaccharidosis type VI (MPS VI, Maroteaux-Lamy 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
- 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 |

## Specimen Stability Information

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

## Clinical & Interpretive

### Clinical Information

Mucopolysaccharidosis VI (MPS VI; Maroteaux-Lamy syndrome) is an autosomal recessive lysosomal disorder caused by the deficiency of N-acetylgalactosamine 4-sulfatase, also known as arylsulfatase B (ARSB) leading to the accumulation of dermatan sulfate. Clinical features and severity of symptoms are widely variable, but typically include short stature, dysostosis multiplex, and degenerative joint disease. Other clinical features may include coarse facial features, hepatosplenomegaly, corneal clouding, and cardiac disease. Cognitive abilities are generally unaffected. Rapidly progressing forms have an early onset of symptoms, significantly elevated glycosaminoglycans (GAGs), and can lead to death before the second or third decades. A more slowly progressing form has a later onset, milder skeletal manifestations, smaller elevations of GAGs, and typically a longer lifespan. Treatment options include hematopoietic stem cell transplantation and/or enzyme replacement therapy.

The differential diagnosis of ARSB deficiency should include multiple sulfatase deficiency and mucolipidosis II (I-Cell disease), however both conditions often present with developmental delays that make them clinically different from MPS VI. The symptoms of MSD mimic metachromatic leukodystrophy (MLD) as well as the mucopolysaccharidoses and can include developmental delay, neurologic regression, dysmorphic facies, dysostosis multiplex, organomegaly,

ichthyosis, and chondroplasia punctata. If MSD is suspected, testing of an additional sulfatase enzyme, such as arylsulfatase A (ARSAW/ Arylsulfatase A, Leukocytes) in MLD, can help determine if multiple sulfatases are deficient. I-cell disease is characterized by congenital or early infantile manifestations including coarse facial features, short stature, skeletal anomalies, cardio- and hepatomegaly, and developmental delays. This is a progressive disorder leading to early death. Additional testing including hydrolase enzymes in serum, such as hexosaminidase A in Tay-Sach disease (NAGS/ Hexosaminidase A and Total Hexosaminidase, Serum) is recommended if a diagnosis of I-cell disease is suspected.

A diagnostic workup for MPS includes glycosaminoglycan determination in urine (MPSQU / Mucopolysaccharides Quantitative, Random, Urine) or blood (MPSBS / Mucopolysaccharidosis, Blood Spot, or MPSER / Mucopolysaccharidosis, Serum) and molecular genetic analysis of the relevant gene. For MPS VI, molecular analysis of the *ARSB* gene (CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies; specify Gene List ID: IEMCP-QQF7DP) allows for detection of disease-causing variant in affected patients and subsequent carrier detection in relatives.

## Reference Values

>0.34 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 11, 2025. https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=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 11, 2025. Available at :https://ommbid.mhmedical.com/content.aspx?bookid=2709&sectionid=225546905
3. D'Avanzo F, Zanetti A, De Filippis C, Tomanin R. Mucopolysaccharidosis Type VI, An updated overview of the disease.

Int J Mol Sci. 2021;22(24):13456. Published 2021 Dec 15. doi:10.3390/ijms222413456

## 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 |
|---|---|---|
| ARSBW | Arylsulfatase B, WBC | 24094-5 |

| Result ID | Test Result Name | Result LOINC® Value |
|---|---|---|
| BG761 | Reason for Referral | 42349-1 |
| 616835 | Arylsulfatase B | 24094-5 |
| 618450 | Interpretation | 59462-2 |
| 618449 | Reviewed By | 18771-6 |