MAYO CLINIC LABORATORIES

Test Definition: ARSBB

Arylsulfatase B, Blood Spot

Overview

Useful For

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

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

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
MPSBS	Mucopolysaccharidosis, BS	Yes	No

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.

Testing Algorithm

If result is normal, testing is complete.

If result indicates mucopolysaccharidosis VI, quantitation of heparan sulfate, dermatan sulfate and keratan sulfate may be performed at an additional charge.

Special Instructions

- Informed Consent for Genetic Testing
- Biochemical Genetics Patient Information
- Blood Spot Collection Card-Spanish Instructions
- Blood Spot Collection Card-Chinese Instructions
- Informed Consent for Genetic Testing (Spanish)
- Blood Spot Collection Instructions

Method Name

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

NY State Available

Yes

Specimen

Specimen Type

Whole blood

Necessary Information

1. Patient's age is required

2. Reason for testing is required

Specimen Required

Submit only 1 of the following specimen types:

Preferred:

Specimen Type: Blood spot

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Blood Spot Collection Card

Acceptable: Whatman Protein Saver 903 Paper, PerkinElmer 226 filter paper, Munktell filter paper, or blood collected in tubes containing ACD or EDTA and dried on filter paper.

Specimen Volume: 2 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see How to Collect a Dried Blood Spot Sample.
2. At least 2 spots should be complete (ie, unpunched).
3. Let blood dry on filter paper at room temperature in a horizontal position for a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. Keep specimen dry.

Specimen Stability Information: Refrigerated (preferred) 60 days/Ambient 7 days/Frozen 60 days

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions.
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777).
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800).

Acceptable:

Specimen Type: Whole Blood

Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Yellow top (ACD)

Specimen Volume: 2 mL

Collection Instructions: Send whole blood specimen in original tube**. Do not aliquot.**

Specimen Stability Information: Refrigerate (preferred) 7 days/Ambient 48 hours

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

Blood spot: 1; Whole blood: 0.5 mL

Reject Due To

Blood spot specimen that shows serum rings or has multiple layers	Reject
Insufficient specimen	Reject
Unapproved filter papers	Reject

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Whole blood	Varies		

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 variants in affected patients and subsequent carrier detection in relatives.

Reference Values

>0.90 nmol/mL/h

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 genetic 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
One dried blood spot sample (DBS) is incubated with a mix of 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. A second DBS sample is incubated with a mix of substrate and IS for acetyl-CoA:alpha-glucosaminide N-acetyltransferase; and a third DBS sample with a mix of substrate and IS for palmitoyl-protein thioesterase 1. Following overnight incubation, the samples are combined, extracted by liquid-liquid extraction, and analyzed by tandem mass spectrometry.(Unpublished Mayo method)

PDF Report
No

Day(s) Performed
Thursday

Report Available
3 to 9 days

Specimen Retention Time
1 year

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
83864 (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
ARSBB	Arylsulfatase B, BS	55912-0

Result ID	Test Result Name	Result LOINC® Value
BG745	Reason for Referral	42349-1

616834	Arylsulfatase B	55912-0
618415	Interpretation	59462-2
618414	Reviewed By	18771-6