

Patient ID SA00176087	Patient Name TESTING, ARSAB_ABNORMAL	Birth Date 2004-02-03	Sex F	Age 21
Order Number SA00176087	Client Order Number SA00176087	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 16 Jun 2025 00:00		

Arylsulfatase A, BS

Interpretation

1 MCR

POSITIVE In this blood spot sample, the activity of arylsulfatase A was reduced. This result can indicate either a diagnosis of Metachromatic Leukodystrophy (MLD), reflect a genotype that confers reduced ARSA activity but not disease ("pseudodeficiency"), or a diagnosis of (ultrare) Multiple Sulfatase deficiency or Saposin B deficiency. We recommend follow up investigations to include arylsulfatase A activity in leukocytes (MCL test ARSAW), urine sulfatides (MCL test CTSU), urine glycosaminoglycans (MCL test MPSQU), and comprehensive molecular genetic analysis of the ARSA gene (MCL test CGPH; gene list ID: IEMCP-WHFH2K). If negative, consider molecular genetic analysis of SUMF1 and PSAP (MCL test CGPH; gene list ID: IEMCP-J13HD9). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-

800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Flow Injection Analysis-Tandem Mass Spectrometry (FIA-MS/MS)

Arylsulfatase A

MCR

0.075 nmol/mL/h
Reference Value
≥0.100

Reviewed By

MCR

Amy Breitlow

Received: 16 Jun 2025 09:29

Reported: 16 Jun 2025 09:33

Laboratory Notes

- 1** This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292