

Arylsulfatase A, Blood Spot

Test ID: ARSAB

Useful for: Enzymatic test for detection of arylsulfatase A deficiency

This test is not suitable for carrier detection.

Ordering Guidance:

This test primarily serves as a second-tier assay to be used by newborn screening programs to screen for metachromatic leukodystrophy when first tier sulfatide analysis is abnormal. The test can also be used to screen patients at risk for metachromatic leukodystrophy.

Method:

Flow Injection Analysis Tandem Mass Spectrometry (MS/MS)

Reference Values:

Normal > or = 0.100 mmol/mL/hr

Specimen Requirements:

Supplies:	Card-Blood Spot Collection (Filter Paper) (T493)		
Preferred:	Card-Blood Spot Collection (Filter Paper)		
Acceptable:	PerkinElmer 226 filter paper, Munktell filter paper, Whatman protein Saver 903 paper, local newborn screening card, or blood collected in tubes containing EDTA (preferred) or heparin and dried on filter paper		
Specimen Volume:	2 Blood spots		
Collection Instructions:	 Completely fill at least 2 circles on the filter paper card (approximately 100 microliters blood per circle). Let blood dry on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours. Do not expose specimen to heat or direct sunlight. Do not stack wet specimens. Keep specimen dry and freeze (with desiccant if necessary). 		

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Whole blood	Frozen (preferred)	28 days	FILTER PAPER
	Refrigerated	14 days	FILTER PAPER

Cautions:

This test is not reliable in identifying carriers.

Arylsulfatase A (ARSA) is also deficient in individuals with multiple sulfatase deficiency and possibly in saposin B deficiency.

Specimens not immediately refrigerated or frozen after drying may yield falsely low ARSA activity results.

This disorder is distinct from conditions caused by deficiencies of aryIsulfatase B (Maroteaux-Lamy disease) and aryIsulfatase C (steroid sulfatase deficiency).

CPT Code: 82657

Day(s) Performed: Monday, Thursday

Report Available: 3 to 6 days

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.