Lab Dept: Chemistry

Test Name: ARYLSULFATASE A, LEUKOCYTES

**General Information**

Lab Order Codes: ARYL

Synonyms: Metachromic Leukodystrophy; Mucolipidoses, Types II and III; ARS-A (Arylsulfatase A); WBC Aryl Sulfatase A

CPT Codes: 82657 – Enzyme activity in blood cells, cultured cells, or tissue, not elsewhere specified; nonradioactive substrate

Test Includes: Arylsulfatase A, Leukocyte level reported in nmol/h/mg.

**Logistics**

Test Indications: Leukocyte assay is the preferred test to order first to rule out metachromatic leukodystrophy. Not reliable in identifying carriers due both to analytical variation and unusual genetic variants. The urine assay should be used in confirming leukocyte results.

Lab Testing Sections: Chemistry - Sendouts

Referred to: Mayo Medical Laboratories (MML Test: ARSAW)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (Specimen must be received by reference lab within 96 hours of collection and must be received 1 day prior to assay day for processing)

Turnaround Time: 8 – 15 days; test set up Tuesday

Special Instructions: Specimen must arrive within 48 hours of draw. Obtain special collection tube from the laboratory.

**Specimen**

Specimen Type: Whole blood

Container: Yellow top (ACD Solution B) tube available from laboratory

Alternate: Yellow top (ACD Solution A)
**Draw Volume:** 6 mL (Minimum: 5 mL) ACD Whole blood

**Processed Volume:** Same as Draw Volume

**Collection:** Routine blood collection

**Special Processing:** Lab Staff: **Do Not** process specimen, leave in original draw container. Refrigerate specimen. **Do Not** transfer blood to other containers. Forward promptly at refrigerated temperatures.

**Patient Preparation:** None

**Sample Rejection:** Frozen or warm specimens; specimens other than ACD; gross hemolysis; mislabeled or unlabeled specimens; specimen older than 96 hours

**Interpretive**

**Reference Range:** > or = 62 nmol/h/mg

Interpretation: Decreased enzyme levels indicate an individual affected with metachromatic leukodystrophy (MLD). Note that individuals with pseudoarylsulfatase A deficiency can have results in this range, but are otherwise unaffected with MLD.

Abnormal results should be confirmed using CTSA/Ceramide Trihexosides and Sulfatides, Urine. If molecular confirmation is desired, consider molecular genetic testing ARSA Gene, Full Gene Analysis.

**Note:** Results from this assay do not reflect carrier status because of individual variation of arylsulfatase A enzyme levels. Low normal values may be due to the presence of pseudodeficiency gene or carrier gene. Patients with these depressed levels may be phenotypically normal.

**Critical Values:** N/A

**Limitations:** This test may not be reliable in identifying carriers due both to analytical variation and unusual genetic variants.

Individuals with psuedodeficiency of arylsulfatase A may have decreased enzyme activity and are not clinically affected with metachromatic leukodystrophy.

Arylsulfatase A is also deficient in individuals with multiple sulfatase deficiency.

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

**Methodology:** Colorimetric Enzyme Assay
References: Mayo Medical Laboratories January 2018

Updates:
2/15/2011: Change in reporting units. Previously reported as U/10^{10} cells.