MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
TESTINGRNV,ARSAS	SA00047977	27	M	SA00047977
Ordering Phys				**DOB** 06/15/1985
Client Order # SA00047977	**Account Information**			**Report Notes**
Collected 07/26/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 08/29/2012 10:08				

Test	Flag	Results	Unit	Reference Value	Perform Site*
ARSA Gene, Full Gene Analysis			REPORTED 08/02/2012 09:38		
Specimen		Blood			MCR
Specimen ID		1038440			MCR
Order Date		26 Jul 2012 09:25			MCR
Reason For Referral					MCR

Patient reported to have features suggestive of metachromatic leukodystrophy (MLD). Test for the presence of mutations within the ARSA gene.

Method MCR

Bi-directional sequence analysis was used performed to test for the presence of a mutations in all coding regions and intron/exon boundaries of the ARSA gene. Mutation nomenclature is based on GenBank accession number; NM_000487.4.

Result MCR

A mutation was NOT detected.

Interpretation MCR

This result decreases the likelihood but does not completely exclude a diagnosis of arylsulfatase A deficiency. We predict that there may be disease causing mutations in the ARSA gene not identifiable by the method described (e.g., large deletions/duplications, promoter mutations, or deep intronic mutations).

In addition, this analysis does not exclude a diagnosis of metachromatic leukodystrophy due to SAP b deficiency. If indicated, consider molecular analysis of the PSAP gene. This result should be interpreted in the context of clinical findings, family history, and other laboratory testing (e.g. arylsulfatase A analysis in leukocytes and urine).

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered. Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. Laboratory developed test.					
Reviewed By		Devin Oglesbee PhD			MCR
Release Date		02 Aug 2012 09:36			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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