MAYO CLINIC
Mayo Medical Laboratories

LABORATORY SERVICE REPORT

1-800-533-1710

PATIENT NAME	PATIENT NUMBER	AGE	SEX	ACCESSION #
TESTING, VLCMS	L3MRNG9158255	34	F	G9158255

ORDERING PHYSICIAN	CLIENT ORDER #	ACCOUNT #
		LIAISONS

COLLECTION DATE TIME	RECEIVED DATE TIME	REPORT PRINTED DATE TIME	SPECIMEN INFORMATION
10/20/10 10:12 A	10/20/10 10:12 A	11/16/10 10:26 A	DATE OF BIRTH:

Test Client
Attn: Mayo Liaisons
200 First Street SW
Rochester, MN 55905
507-284-8202

TEST REQUESTED	HI LO		REF RANGE	PERFORM SITE *
VLCAD Deficiency,Full Gene Analysis				
Specimen		**Blood**		**MCR**
Specimen ID		**1034804**		**MCR**
Order Date		**20 Oct 2010 10:25**		**MCR**
Reason for Referral				**MCR**

Patient reported to have features suggestive of very long chain acyl-CoA dehydrogenase (VLCAD) deficiency. Test for the presence of mutations in the ACADVL gene.

Method **MCR**

DNA sequence analysis was used to test for the presence of a mutation in all 20 exons of the ACADVL gene. Mutation nomenclature is based on GenBank accession number; NM_000018.2.

Result **MCR**

A mutation was NOT detected.

Interpretation **MCR**

This result decreases the likelihood but does not completely exclude carrier or affected status for very long chain acyl-CoA dehydrogenase (VLCAD) deficiency. We predict that a small percentage of individuals with VLCAD deficiency may have mutations that are not identifiable by the method described (e.g., large deletions/duplications, promoter mutations, or deep intronic mutations).

If testing was performed to rule out a diagnosis of VLCAD deficiency, this result should be interpreted in the context of clinical findings, family history, and other laboratory testing such as plasma acylcarnitines and fatty acid oxidation probe assay in skin fibroblasts.

If testing was performed because of a family history of VLCAD deficiency, genetic testing of an affected individual should be considered. Identification of the mutations in

** Perform Site Legend on last page of report*

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this family would allow for more accurate risk assessment of at risk individuals.
A genetic consultation may be of benefit.
A list of common polymorphisms identified for this patient is 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 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.
This test was developed and its performance characteristics determined by Laboratory Medicine and Pathology, Mayo Clinic. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Extraction Performed?	**Yes**	**MCR**
Reviewed By		**MCR**
Michelle Dawn Fjerstad		
Release Date	**20 Oct 2010 10:30**	**MCR**

*** PERFORMING SITE**

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First Street SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.

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TESTING, VLCMS	Final	10/20/10 10:12 A