MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLEREPORT,SCADK N	SA00058858	46	F	SA00058858
Ordering Phys CLIENT,CLIENT				**DOB** 06/10/1966
Client Order # SA00058858	**Account Information**			**Report Notes**
Collected 06/06/2013 00:00	C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			
Printed 06/11/2013 16:11				

Test	Flag	Results	Unit	Reference Value	Perform Site*
SCAD Known Mutation					
RECEIVED: 06/07/2013 16:03 **REPORTED:** 06/11/2013 08:06					
Specimen		Blood			MCR
Specimen ID		1062125			MCR
Order Date		10 Jun 2013 09:17			MCR
Reason For Referral					MCR
Family history of Short-Chain Acyl-CoA Dehydrogenase (SCAD) deficiency. Test for the presence of familial alteration(s) within the ACADS gene.					
Method					MCR
DNA sequence analysis was used to test for the presence of the p.W177R (c.529T>C) and p.A192V (c.575C>T) alterations in exon 5 of the ACADS gene (GenBank accession number; NM_000017). Analysis for these specific alterations was performed because they are known to be present in a family member.					
Results					MCR
The p.W177R and p.A192V alterations were NOT detected.					
Interpretation					MCR
Absence of the mutation(s) previously identified for an family member indicates that this individual is at no greater risk than someone in the general population for being a carrier of, or affected with, SCAD deficiency.					
This assay does not rule out the presence of other mutations within this gene or within other genes that are associated with metabolic disease. Any error in the diagnosis or in the pedigree provided to us, including false paternity, could lead to an erroneous interpretation of results.					
A genetic consult 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 provided is inaccurate or incomplete.					

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

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Rare polymorphisms exist that could lead to false negative or false positive results. If results obtained do not match the clinical and biochemical 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:					MCR
Emily Christine Lauer					
Release Date		11 Jun 2013 08:04			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director: