MAYO CLINIC
Mayo Medical Laboratories

NEW TEST ANNOUNCEMENT
NOTIFICATION DATE: November 16, 2010
EFFECTIVE DATE: November 19, 2010

Very Long Chain Acyl-CoA Dehydrogenase Deficiency, Full Gene Analysis
#60036

USEFUL FOR:
- Confirmation of a diagnosis of very long chain acyl-CoA dehydrogenase (VLCAD) deficiency
- Carrier screening in cases where there is a family history of VLCAD deficiency, but an affected individual is not available for testing or disease-causing mutations have not been identified

METHODOLOGY: Polymerase Chain Reaction (PCR)/DNA Sequencing Analysis

REFERENCE VALUES: An interpretive report will be provided.

SPECIMEN REQUIREMENTS: **Blood:** 3 mL of whole blood in a Lavender-top (EDTA) tube or yellow-top (ACD) tube. Specimen must arrive within 96 hours of collection.
NOTE: **Alternate:** 5 blood spots on "Whatman Protein Saver 903 Paper"

CAUTIONS:
- A small percentage of individuals who are carriers or have a diagnosis of VLCAD deficiency may have a mutation that is not identified by this method (eg, large genomic deletions, promoter mutations). The absence of a mutation(s), therefore, does not eliminate the possibility of positive carrier status or the diagnosis of VLCAD deficiency. For carrier testing, it is important to first document the presence of an *ACADVL* gene mutation in an affected family member.
- In some cases, DNA alterations of undetermined significance may be identified.
- Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.
- A previous bone marrow transplant from an allogenic donor will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.
- Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

LIST FEE: $1194.85
CPT CODE:
83891-Isolation or extraction of highly purified nucleic acid
83898 x 13-Amplification, target, each nucleic acid sequence
83909 x 26-Separation and identification by high-resolution technique
83912-Interpretation and report

ANALYTIC TIME: 5 days **DAY(S) SET-UP:** Tuesdays, 10 AM

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or
Marvin H. Anderson, Jr., MML Laboratory Technologist Resource Coordinator
Telephone: 800-533-1710