ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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THIS IS NOT A TEST REQUEST FORM.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR VERY LONG CHAIN ACYL-CoA DEHYDROGENASE (VLCAD) DEFICIENCY

Patient Name _____ Date of Birth _____ Sex ☐ F ☐ M

Physician _____ Physician Phone _____

Practice Specialty _____ Physician Fax _____

Genetic Counselor _____ Counselor Phone _____

Patient's Ethnicity (check all that apply)

☐ African-American ☐ Asian ☐ Hispanic ☐ Native American
☐ Ashkenazi Jewish ☐ Caucasian ☐ Middle Eastern ☐ Other: _____

Did the patient have an abnormal newborn screen? ☐ No ☐ Yes ☐ Unknown

Does the patient have symptoms? ☐ No ☐ Yes (check all that apply)

☐ Cardiomyopathy
☐ Coma
☐ Encephalopathy
☐ Hepatomegaly
☐ Hypoglycemia
☐ Lethargy
☐ Liver failure
☐ Reye-like syndrome
☐ Rhabdomyolysis
☐ Other symptom(s): _____

Laboratory Findings:

Plasma acylcarnitine profile:.. ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown
Urine organic acids: ..... ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown
Plasma carnitine (without supplements):
Free/total:..... ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown

Is there any relevant family history? ☐ No ☐ Yes ☐ Unknown
If yes, attach a pedigree or specify the relative's relationship to the patient: _____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown
If yes, attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Check the test you intend to order.

☐ **2004212 Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing and Deletion/Duplication**: Clinical sensitivity may be as high as 95%; sequencing and deletion/duplication analysis of the ACADVL coding regions and intron/exon boundaries.

☐ **2002001 Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Sequencing:** Clinical sensitivity 80–90%; sequencing of the ACADVL coding regions and intron/exon boundaries.

☐ **2004208 Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) Deletion/Duplication:** Clinical sensitivity may be as high as 10%; deletion/duplication analysis of the ACADVL coding regions.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for a mutation previously identified in a family member; a copy of relative's lab result is REQUIRED.

Master Label

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141