

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform VLCAD deficiency testing.
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 ____/____/____ Gender F M

Physician _____ Physician Phone (____) _____ Practice Specialty _____

Genetic Counselor _____ Counselor Phone (____) _____

PATIENT ETHNICITY (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

SYMPTOMS of VLCAD? No Yes Unknown

If yes, check all that apply

- | | |
|---|--|
| <input type="checkbox"/> Hypoglycemia | <input type="checkbox"/> Hepatomegaly |
| <input type="checkbox"/> Cardiomyopathy | <input type="checkbox"/> Liver failure |
| <input type="checkbox"/> Encephalopathy | <input type="checkbox"/> Lethargy |
| <input type="checkbox"/> Reye-like syndrome | <input type="checkbox"/> Coma |
| <input type="checkbox"/> Rhabdomyolysis | <input type="checkbox"/> Other _____ |

ABNORMAL NEWBORN SCREEN? No Yes Unknown

LABORATORY FINDINGS

- | | | | | |
|------------------------------|---------------------------------|-----------------------------------|--|----------------------------------|
| Plasma acylcarnitine profile | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
| Urine organic acids | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |

Plasma carnitine (**without supplements**):

- | | | | | |
|---------------|---------------------------------|-----------------------------------|--|----------------------------------|
| Free / Total: | <input type="checkbox"/> Normal | <input type="checkbox"/> Abnormal | <input type="checkbox"/> Not performed | <input type="checkbox"/> Unknown |
|---------------|---------------------------------|-----------------------------------|--|----------------------------------|

FAMILY HISTORY OF VLCAD DEFICIENCY? No Yes Unknown

If yes, describe relationship(s) to the patient _____

Has DNA testing for VLCAD been performed for these family member(s)? No Yes Unknown

If yes, please attach a copy of the laboratory result (REQUIRED for familial mutation testing)

Circle the VLCAD 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: Targeted sequencing for a *ACADVL* mutation previously identified in a family member.

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

Master Label