ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

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

500 Chipeta Way
Salt Lake City, Utah 84108-1221
Phone: 801-583-2787 | toll free: 800-242-2787
Fax: 801-584-5249 | aruplab.com

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 MEDIUM CHAIN ACYL-CoA DEHYDROGENASE (MCAD) DEFICIENCY

Patient Name: _____ **Date of Birth:** _____ **Sex**: ☐ Female ☐ Male

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: _____

Clinical Diagnosis/Reason for Referral: _____

Did the patient have an abnormal newborn screen? ☐ No ☐ Yes ☐ Not Performed ☐ Unknown

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

☐ Coma ☐ Hepatomegaly ☐ Lethargy ☐ Reye-like syndrome ☐ Vomiting

☐ Encephalopathy ☐ Hypoglycemia ☐ Liver failure ☐ Seizures

☐ Other symptom(s): _____

Laboratory Findings

Plasma acylcarnitine profile: ..... ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown

Urine organic acids: ..... ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown

Urine acylglycine: ..... ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown

Plasma carnitine (without supplements): Free/total: ..... ☐ Normal ☐ Abnormal ☐ Not performed ☐ Unknown

Is there any relevant family history of MCAD deficiency? ☐ No ☐ Yes ☐ Unknown

Attach a pedigree or specify the relationship of family member(s) to the patient: _____

Has DNA testing been performed for the family member(s)? ☐ No ☐ Yes ☐ Unknown

If yes, list the MCAD mutations identified in the family member: _____

The relative is: ☐ a healthy carrier ☐ affected with MCAD

Attach a copy of the relative's DNA laboratory result (REQUIRED for familial mutation testing).

Check the test you intend to order.

☐ **0051205 MCAD Deficiency (*ACADM*) 2 Mutations:** Testing for two common *ACADM* gene mutations A985G (K304E) and T199C (Y42H); clinical sensitivity is 75%.

☐ **0051758 MCAD Deficiency (*ACADM*) Sequencing:** Sequencing *ACADM* coding regions and intron/exon boundaries; clinical sensitivity ~95%. Diagnostic testing for patients without 2 identifiable mutations using the MCAD (*ACADM*) 2 Mutation test.

☐ **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