ARUP LABORATORIES

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 complete and submit with the test request form or electronic packing list.

MEDIUM CHAIN ACYL-CoA DEHYDROGENASE (MCAD) DEFICIENCY PATIENT HISTORY FORM

Patient Name: _____ **Date of Birth:** _____

Sex Assigned at Birth: ☐Female ☐Male ☐Intersex **Gender Identity (optional):** ☐Female ☐Male ☐_____

Ordering Provider: _____ **Provider's Phone:** _____

Practice Specialty: _____ **Provider's Fax:** _____

Genetic Counselor: _____ **Counselor's Phone:** _____

Patient's Ethnicity/Ancestry (check all that apply)

☐ African American/Black ☐ Asian ☐ Hispanic ☐ White ☐ Other: _____

List country of origin (if known): _____

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

Check the test you intend to order.

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

☐ **3001851 Fatty Acid Oxidation Disorders Panel, Sequencing:** Preferred molecular test to confirm or rule out a diagnosis of a fatty acid oxidation disorder following clinical and/or biochemical presentations.

☐ **2001961 Familial Mutation, Targeted Sequencing:** Tests for sequence variant(s) previously identified in the family. Contact an ARUP genetic counselor at 800-242-2787 ext 2141 prior to test submission to discuss requirements.

Master Label

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