

**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

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

