

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform MCAD deficiency testing.
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's Name _____ **Date of Birth** ____/____/____ **Gender** F M
Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____
Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)
 African American Ashkenazi Jewish Asian Caucasian
 Hispanic Middle Eastern Native American Other _____

NEWBORN SCREEN Normal Abnormal Not performed Unknown

Does the patient have SYMPTOMS? No Yes Unknown If yes, check all that apply
 Hypoglycemia Hepatomegaly Liver failure
 Vomiting Reye-like syndrome Encephalopathy
 Lethargy Seizures Coma Other _____

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

Does the patient have a FAMILY HISTORY OF MCAD DEFICIENCY? No Yes
If yes, attach a PEDIGREE or specify the **RELATIONSHIP** of family member(s) to the patient. _____
Is the relative a healthy carrier or affected with MCAD?
What are the MCAD mutations in the family member? _____

Circle the MCAD 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:** A copy of the relative's DNA lab result is **REQUIRED** for this test.

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

Master Label