ARUP LABORATORIES

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 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.

Master Label

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