NEWBORN SCREENING PROGRAM
New York State Department of Health
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INHERITED METABOLIC DISORDER–FATTY ACID OXIDATION - DIAGNOSIS FORM

Dear Doctor:
Please complete this form in its entirety and return it to the Newborn Screening Program as soon as possible.
Attach Clinical Laboratory results including any available mutation analysis.
Your response is required, as specified in Title 10 New York Code of Rules and Regulations subpart 69-1.7c.

NEWBORN INFORMATION

Name at birth: _____
AKA: _____

Single Birth ☐ Twin A ☐ Twin B ☐ Other _____
Mother's name: _____
Date of Birth: _____

Gender: Male ☐ Female ☐
Hospital of birth: _____
Medical Record #: _____

CARN DEFICIENCY

CUD01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
CUD10 [ ] Disease, Carnitine uptake defect (CUD)
CUD29 [ ] Disease, not on NBS panel. Specify: _____
CUD30 [ ] Inconclusive/possible (work-up in progress), CUD
CUD40 [ ] No disease
CUD41 [ ] No disease, transient deficiency due to prematurity/TPN
CUD49 [ ] No disease, polymorphisms only
CUD71 [ ] Other, maternal disease or medication

SCADD

SCAD0 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
SCAD10 [ ] Disease, Short-chain acyl-CoA dehydrogenase (SCAD) deficiency
SCAD11 [ ] Disease, Isobutyryl-CoA dehydrogenase (IBDH) deficiency-isobutyrylglycinuria (IBG)
SCAD12 [ ] Disease, Ethylmalonic encephalopathy (EMA)
SCAD29 [ ] Disease, not on NBS panel. Specify: _____
SCAD30 [ ] Inconclusive/possible (work-up in progress), SCADD/IBDH/EMA
SCAD40 [ ] No disease
SCAD41 [ ] No disease, transient deficiency due to prematurity/TPN
SCAD71 [ ] Other, maternal disease or medication

MCADD/MADD

MCAD01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
MCAD10 [ ] Disease, Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
MCAD11 [ ] Disease, Medium-chain 3-keto acyl-CoA thiolase (MCKAT) deficiency
MCAD12 [ ] Disease, Multiple acyl-CoA dehydrogenase (MAD) deficiency – glutaric acidemia type 2
MCAD29 [ ] Disease, not on NBS panel. Specify: _____
MCAD30 [ ] Inconclusive/possible (work-up in progress), MCADD/MCKAT/MADD
MCAD40 [ ] No disease
MCAD41 [ ] No disease, transient deficiency due to prematurity/TPN

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MCAD49 [ ] No disease, polymorphisms only
MCAD71 [ ] Other, maternal disease or medication

VLCADD

VLCA01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
VLCA10 [ ] Disease, Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
VLCA29 [ ] Disease, not on NBS panel. Specify: _____
VLCA30 [ ] Inconclusive/possible (work-up in progress), VLCAD
VLCA40 [ ] No disease
VLCA41 [ ] No disease, transient deficiency due to prematurity/TPN
VLCA45 [ ] No disease, Carrier
VLCA71 [ ] Other, maternal disease or medication

LCHADD/TFP

LCHA01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
LCHA10 [ ] Disease, Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
LCHA11 [ ] Disease, Trifunctional protein (TFP) deficiency
LCHA29 [ ] Disease, not on NBS panel. Specify: _____
LCHA30 [ ] Inconclusive/possible (work-up in progress), LCHAD/TFP
LCHA40 [ ] No disease
LCHA41 [ ] No disease, transient elevation due to prematurity/TPN
LCHA71 [ ] Other, maternal disease or medication

CPT-II/CAT

CPT201 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
CPT210 [ ] Disease, Carnitine palmitoyltransferase II (CPT2) deficiency
CPT211 [ ] Disease, Carnitine/acylcarnitine translocase (CACT) deficiency
CPT229 [ ] Disease, not on NBS panel. Specify: _____
CPT230 [ ] Inconclusive/possible (work-up in progress), CPT2/CACT
CPT240 [ ] No disease
CPT241 [ ] No disease, transient elevation due to prematurity/TPN
CPT271 [ ] Other, maternal disease or medication

2,4-DI

24DI01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
24DI10 [ ] Disease, 2,4-Dienoyl-CoA (2,4Di) reductase deficiency
24DI29 [ ] Disease, not on NBS panel. Specify: _____
24DI30 [ ] Inconclusive/possible (work-up in progress), 2,4Di
24DI40 [ ] No disease
24DI41 [ ] No disease, transient elevation due to prematurity/TPN
24DI71 [ ] Other, maternal disease or medication

CPT-1

CPT101 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
CPT110 [ ] Disease, Carnitine palmitoyltransferase 1 (CPT1) deficiency
CPT129 [ ] Disease, not on NBS panel. Specify: _____
CPT130 [ ] Inconclusive/possible (work-up in progress), Possible disease, CPT1
CPT140 [ ] No Disease
CPT141 [ ] No Disease, transient elevation due to prematurity/TPN
CPT171 [ ] Other, maternal disease or medication

M/SCHAD

MSCH01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below.
MSCH10 [ ] Disease, Medium/short-chain hydroxyl CoA dehydrogenase (M/SCHAD) deficiency
MSCH29 [ ] Disease, not on NBS panel. Specify: _____
MSCH30 [ ] Inconclusive/possible (work-up in progress), M/SCHAD
MSCH40 [ ] No disease
MSCH41 [ ] No disease, transient elevation due to prematurity/TPN
MSCH71 [ ] Other, maternal disease or medication

Was this newborn previously known to be at increased risk for this disorder?
[ ] No [ ] Yes, family history [ ] Yes, prenatal testing [ ] Yes, preconception testing

COMMENTS:_____
PHYSICIAN'S SIGNATURE: _____**DATE:**_____
PRINT NAME: _____