NEWBORN SCREENING PROGRAM
New York State Department of Health
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Albany, NY 12208
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Website: http://www.wadsworth.org/newborn/

INHERITED METABOLIC DISORDER–ORGANIC ACID - 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 #: _____

Diagnosis Date:_____

PA/MMA

PAMM01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below
PAMM10 [ ] Disease, Propionyl-CoA carboxylase deficiency – propionic acidemia (PA)
PAMM11 [ ] Disease, Methylmalonyl-CoA mutase deficiency (mut0 or mut-)
PAMM12 [ ] Disease, Cobalamin A/B deficiency
PAMM13 [ ] Disease, Cobalamin C/D/F deficiency
PAMM14 [ ] Disease, Transcobalamin II deficiency
PAMM15 [ ] Disease, Vitamin B12 deficiency
PAMM29 [ ] Disease, not on NBS panel. Specify:_____
PAMM30 [ ] **Inconclusive**/possible (work-up in progress), PA/MMA
PAMM40 [ ] No disease
PAMM41 [ ] No disease, transient elevation due to prematurity/TPN
PAMM71 [ ] Other, maternal disease or medication

IVA

IVA01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below
IVA10 [ ] Disease, Isovaleryl CoA dehydrogenase deficiency – isovaleric acidemia (IVA)
IVA11 [ ] Disease, 2-Methylbutyrylglycinuria (2MBG) – 2-methylbutyryl-CoA dehydrogenase (2MBCD) deficiency-short/branched chain acyl-CoA dehydrogenase (SBCAD) Deficiency
IVA29 [ ] Disease, not on NBS panel. Specify:_____
IVA30 [ ] **Inconclusive**/possible (work-up in progress), IVA
IVA40 [ ] No disease
IVA41 [ ] No disease, transient elevation due to prematurity/TPN
IVA71 [ ] Other, maternal disease or medication

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GA1

GA101 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below
GA110 [ ] Disease, Glutaryl-CoA dehydrogenase deficiency-glutaric aciduria (GA-1)
GA129 [ ] Disease, not on NBS panel. Specify:_____
GA130 [ ] **Inconclusive**/possible (work-up in progress), GA-1
GA140 [ ] No disease
GA141 [ ] No disease, transient elevation due to prematurity/TPN
GA171 [ ] Other, maternal disease or medication

3MCC/HMG

3MCC01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below
3MCC10 [ ] Disease, 3-Methylcrotonyl-CoA carboxylase (3MCC) deficiency, clinically significant
3MCC11 [ ] Disease, 3-Methylcrotonyl-CoA Carboxylase (3MCC) deficiency, not clinically significant
3MCC12 [ ] Disease, 3-Hydroxy-3-methylglutaryl-CoA lyase (HMG) deficiency
3MCC13 [ ] Disease, β -Ketothiolase (BKT) deficiency
3MCC14 [ ] Disease, 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD) deficiency – 2 - Methyl-3-hydroxybutyric acidemia (2M3HBA)
3MCC15 [ ] Disease, 3-Methylglutaconic aciduria (3MGA)
3MCC16 [ ] Disease, Biotinidase deficiency
3MCC17 [ ] Disease, Holocarboxylase deficiency
3MCC18 [ ] Disease, Biotin deficiency
3MCC29 [ ] Disease, not on NBS panel. Specify:_____
3MCC30 [ ] **Inconclusive**/possible (work-up in progress), 3MCC/HMG/BKT/MCD/MHBD/3MGA
3MCC40 [ ] No disease
3MCC41 [ ] No disease, transient elevation due to prematurity/TPN
3MCC71 [ ] Other, maternal disease or medication

BKT

BKT01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below
BKT10 [ ] Disease, Mitochondrial acetoacetyl-CoA thiolase deficiency-beta-ketothiolase (BKT) deficiency
BKT11 [ ] Disease, 2-Methyl-3-hydroxybutyryl-CoA-dehydrogenase (MHBD) deficiency
BKT29 [ ] Disease, not on NBS panel. Specify:_____
BKT30 [ ] **Inconclusive**/possible (work-up in progress), BKT/MHBD
BKT40 [ ] No disease
BKT41 [ ] No disease, transient elevation due to prematurity/TPN
BKT49 [ ] No disease, polymorphisms only
BKT71 [ ] Other, maternal disease or medication

MA

MA01 [ ] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below
MA10 [ ] Disease, Malonyl-CoA decarboxylase deficiency – malonic aciduria (MA)
MA29 [ ] Disease, not on NBS panel. Specify:_____
MA30 [ ] **Inconclusive**/possible (work-up in progress), MA
MA40 [ ] No disease
MA41 [ ] No disease, transient elevation due to prematurity/TPN
MA71 [ ] 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:_____