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 #: _______________________________

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

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

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

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