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

Dear Doctor:
Please complete this form in its entirety and return it to the Newborn Screening Program as soon as possible.

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 #: _____

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.

MSUD-MS

MSUD01 **[ ]** Expired, If cause of death is known, choose the appropriate diagnosis below.
MSUD10 [ ] Disease, Maple Syrup Urine Disease (MSUD)
MSUD11 [ ] Disease, Hydroxyprolinemia
MSUD29 [ ] Disease, no on NBS panel. Specify:_____
MSUD30 [ ] Inconclusive/possible (work-up in progress), MSUD
MSUD40 [ ] No disease
MSUD41 [ ] No disease, transient elevation due to prematurity/TPN
MSUD71 [ ] Other, maternal disease or medication

HCY-MS

HCY01 [ ] Expired, If cause of death is known, choose the appropriate diagnosis below.
HCY10 [ ] Disease, Homocystinuria (HYC), cystathionine β-synthase deficiency
HCY11 [ ] Disease, Hypermethioninemia (HMET) due to methyladenosyltransferase (MAT) l/lll deficiency
HCY12 [ ] Disease, Hypermethioninemia (HMET) due to guanidinoacetate methyltransferase (GNMT) deficiency
HCY13 [ ] Disease, Hypermethioninemia (HMET) due to adenosylhomocysteine (AdoHcy) hydrolase deficiency
HCY29 [ ] Disease, not on NBS panel. Specify:_____
HCY30 [ ] Inconclusive/possible (work-up in progress), HCY/HMET
HCY40 [ ] No disease
HCY41 [ ] No disease, transient elevation due to prematurity/TPN
HCY71 [ ] Other, maternal disease or medication

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PKU-MS

PKU01 [ ] Expired, If cause of death is known, choose the appropriate diagnosis below.
PKU10 [ ] Disease, Phenylketonuria (PKU) – classical, due to phenylalanine hydroxylase (PAH) deficiency
PKU11 [ ] Disease, Phenylketonuria (PKU) variant
PKU12 [ ] Disease, Hyperphenylalaninemia (HPHE) due to guanine triphosphate cyclohydrolase (GTPCH) deficiency
PKU13 [ ] Disease, Hyperphenylalaninemia (HPHE) due to 6-pyruvoyl tetrahydrobiopterin synthase (PTPS) deficiency
PKU14 [ ] Disease, Hyperphenylalaninemia (HPHE) due to dihydropterdine reductase (DHPR) deficiency
PKU15 [ ] Disease, Hyperphenylalaninemia (HPHE) due to pterin-4 acarbinolamine dehydratase (PCD) deficiency
PKU16 [ ] Disease, Hyperphenylalaninemia (HPHE) not otherwise specified (NOS) clinically significant
PKU29 [ ] Disease, not on NBS panel. Specify:_____
PKU30 [ ] Inconclusive/possible (work-up in progress), PKU/HPHE
PKU40 [ ] No disease
PKU41 [ ] No disease, transient elevation due to prematurity/TPN
PKU42 [ ] No disease, benign hyperphenylalaninemia
PKU71 [ ] Other, maternal disease or medication

Tyrosinemia Type 2,3

TYR201 [ ] Expired, If cause of death is known, choose the appropriate diagnosis below.
TYR210 [ ] Disease, Tyrosinemia Type 2 (oculocutaneous)
TYR211 [ ] Disease, Tyrosinemia Type 3
TYR229 [ ] Disease, not on NBS panel. Specify:_____
TYR230 [ ] Inconclusive/possible (work-up in progress), TYR 2,3
TYR240 [ ] No disease
TYR241 [ ] No disease, transient elevation due to prematurity/TPN
TYR242 [ ] No disease, Transient Tyrosinemia 2,3 of the newborn (TTN)
TYR271 [ ] Other, maternal disease or medication

Tyrosinemia Type 1

TYR101 [ ] Expired, If cause of death is known, choose the appropriate diagnosis below.
TYR110 [ ] Disease, Tyrosinemia Type 1 (hepatorenal)
TYR129 [ ] Disease, not on NBS panel. Specify:_____
TYR130 [ ] Inconclusive/possible (work-up in progress), TYR 1
TYR140 [ ] No disease
TYR141 [ ] No disease, Transient elevation due to prematurity/TPN
TYR142 [ ] No disease, Transient Tyrosinemia Type 1 of the newborn (TTN)
TYR171 [ ] 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:_____**FACILITY/PRACTICE:**_____