NEWBORN SCREENING PROGRAM
New York State Department of Health
Wadsworth Center, David Axelrod Institute
120 New Scotland Avenue
Albany, NY 12208
Phone: (518)473-7552 Fax: (518)474-0405
E-mail: nbsinfo@health.ny.gov

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.
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 69-1.5e.

NEWBORN INFORMATION:

Name at Time of Birth: _____

Other Names (AKA): _____

Single Birth ☐ Twin A ☐ Twin B ☐ Other _____

Mother's Name: _____

Date of Birth: _____

Gender: Male ☐ Female ☐

Hospital of Birth: _____

Medical Record #: _____

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, not on NBS panel. Specify: _____
MSUD30 [ ] Inconclusive, 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, 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) ï 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, 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, Tyrosinemia Type 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, Tyrosinemia Type 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

COMMENTS: _____

PHYSICIAN'S SIGNATURE: _____DATE:_____

PRINT NAME: _____FACILITY/PRACTICE: _____