

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.

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: _____

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) I/III 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 dihydropteridine 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:** _____