

NEWBORN SCREENING PROGRAM
New York State Department of Health
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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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GAI

- 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: _____