

NEWBORN SCREENING PROGRAM
New York State Department of Health
Wadsworth Center, David Axelrod Institute
120 New Scotland Avenue
Albany, NY 12208
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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 69-1.5e.

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 #: _____

PA/MMA

PAMM01 [] Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below

PAMM10 [] Disease, Propionyl-CoA carboxylase deficiency ó 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**, 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 ó isovaleric acidemia (IVA)

IVA11 [] Disease, 2-Methylbutyrylglucineria (2MBG) ó 2-methylbutyryl-CoA dehydrogenase (2MBCD) deficiency-short/branched chain acyl-CoA dehydrogenase (SBCAD) deficiency

IVA29 [] Disease, not on NBS panel. Specify: _____

IVA30 [] **Inconclusive**, IVA

IVA40 [] No disease

IVA41 [] No disease, transient elevation due to prematurity/TPN

IVA71 [] Other, maternal disease or medication

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GA1

- 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**, 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, -Ketothiolase (BKT) deficiency
- 3MCC14 [] Disease, 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase (MHBD) deficiency ó
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**, 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**, 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 ó malonic aciduria (MA)
- MA29 [] Disease, not on NBS panel. Specify: _____
- MA30 [] **Inconclusive**, MA
- MA40 [] No disease
- MA41 [] No disease, transient elevation due to prematurity/TPN
- MA71 [] Other, maternal disease or medication

COMMENTS: _____

PHYSICIAN'S SIGNATURE: _____ **DATE:** _____

PRINT NAME: _____