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 #: _____________________________

Diagnosis Date: ________________________________

<table>
<thead>
<tr>
<th>PA/MMA</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>PAMM01</td>
<td>Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below</td>
</tr>
<tr>
<td>PAMM10</td>
<td>Disease, Propionyl-CoA carboxylase deficiency – propionic acidemia (PA)</td>
</tr>
<tr>
<td>PAMM11</td>
<td>Disease, Methylmalonyl-CoA mutase deficiency (mut0 or mut-)</td>
</tr>
<tr>
<td>PAMM12</td>
<td>Disease, Cobalamin A/B deficiency</td>
</tr>
<tr>
<td>PAMM13</td>
<td>Disease, Cobalamin C/D/F deficiency</td>
</tr>
<tr>
<td>PAMM14</td>
<td>Disease, Transcobalamin II deficiency</td>
</tr>
<tr>
<td>PAMM15</td>
<td>Disease, Vitamin B12 deficiency</td>
</tr>
<tr>
<td>PAMM29</td>
<td>Disease, not on NBS panel. Specify:</td>
</tr>
<tr>
<td>PAMM30</td>
<td>Inconclusive/possible (work-up in progress), PA/MMA</td>
</tr>
<tr>
<td>PAMM40</td>
<td>No disease</td>
</tr>
<tr>
<td>PAMM41</td>
<td>No disease, transient elevation due to prematurity/TPN</td>
</tr>
<tr>
<td>PAMM71</td>
<td>Other, maternal disease or medication</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>IVA</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>IVA01</td>
<td>Expired, no diagnosis. If cause of death is known, choose the appropriate diagnosis below</td>
</tr>
<tr>
<td>IVA10</td>
<td>Disease, Isovaleryl CoA dehydrogenase deficiency – isovaleric acidemia (IVA)</td>
</tr>
<tr>
<td>IVA11</td>
<td>Disease, 2-Methylbutyrylglycinuria (2MBG) – 2-methylbutyryl-CoA dehydrogenase (2MBCD) deficiency-short/branched chain acyl-CoA dehydrogenase (SBCAD) Deficiency</td>
</tr>
<tr>
<td>IVA29</td>
<td>Disease, not on NBS panel. Specify:</td>
</tr>
<tr>
<td>IVA30</td>
<td>Inconclusive/possible (work-up in progress), IVA</td>
</tr>
<tr>
<td>IVA40</td>
<td>No disease</td>
</tr>
<tr>
<td>IVA41</td>
<td>No disease, transient elevation due to prematurity/TPN</td>
</tr>
<tr>
<td>IVA71</td>
<td>Other, maternal disease or medication</td>
</tr>
</tbody>
</table>

LEIO30(continued on back or page 2)
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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/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:________________________________________

Enclosures  LEIO30