NEWBORN SCREENING PROGRAM
New York State Department of Health
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POMPE DISEASE REFERRAL DIAGNOSIS FORM

Please complete this form in its entirety and return it to the Newborn Screening Program as soon as possible. Your response is required, as specified in Title 10 New York Code of Rules and Regulations subpart 69-1.7c. Note: Newborn Screening results do not constitute a diagnosis. Confirmatory testing is required.

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 #: __________________________________

1. Abnormal clinical findings/symptoms?   [ ] Yes    [ ] No

2. If yes, please specify:
   [ ] Hypotonia
   [ ] Cardiomegaly
   [ ] Difficulty feeding
   [ ] Breathing difficulties
   [ ] Delayed developmental milestones
   [ ] Failure to thrive
   [ ] Respiratory infection(s)
   [ ] Hearing loss
   [ ] Ptosis
   [ ] Other, specify __________________________________________

3. Cardiac evaluation:    [ ] Normal    [ ] Abnormal    [ ] Not Done

4. Maternal Ethnicity ________________________  Paternal Ethnicity ________________________
5. Confirmatory testing

<table>
<thead>
<tr>
<th>DATE</th>
<th>TEST</th>
<th>Newborn’s Results</th>
<th>Normal Range</th>
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<tbody>
<tr>
<td></td>
<td>Leukocyte GAA</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Urine Glc₄</td>
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6. Select genotype:

- [ ] 2 disease-causing mutations
- [ ] 1 disease-causing mutation, ≥ 1 VUS
- [ ] 1 disease-causing mutation
- [ ] 2 VUS
- [ ] 1 VUS
- [ ] Pseudodeficiency Allele(s)

7. Select diagnosis:

- [ ] Infantile-onset Pompe disease
- [ ] Pompe disease (asymptomatic)
- [ ] Possible Pompe disease
- [ ] Carrier of Pompe disease

8. 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: ______________________