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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</thead>
<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

Diagnosis Date: ________________

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: ___________________________