NEWBORN SCREENING PROGRAM
New York State Department of Health
Wadsworth Center, David Axelrod Institute, 120 New Scotland Ave.
Albany, NY 12208
Phone: (518)473-7552 Fax: (518) 474-0405
E-mail: nbsinfo@health.state.ny.us
Website: http://www.wadsworth.org/newborn/

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**

DATE	TEST	Newborn's Results	Normal Range
	Leukocyte GAA		
	Urine Glc_4		
	CK		

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:** ___________________________