

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

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

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: _____