
Reporting Title: Pompe Disease 2ND Tier NBS, BS**Performing Location:** Rochester**Ordering Guidance:**

Due to reference range differences, this is the appropriate test for patients less than or equal to 6 weeks of age. For patients greater than 6 weeks of age, order PDBS / Pompe Disease, Blood Spot.

Necessary Information:

1. Birth weight (grams)
2. Time of birth (24-hour time)
3. Gestational age (weeks)

Specimen Requirements:

Supplies: Card-Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Blood Spot Collection Card

Acceptable: PerkinElmer 226 filter paper, Munktell filter paper, Whatman Protein Saver 903 paper, local newborn screening card, or blood collected in tubes containing ACD or EDTA and then spotted and dried on filter paper.

Specimen Volume: 3 Blood spots

Collection Instructions:

1. Completely fill at least 3 circles on the filter paper card (approximately 100 microliters blood per circle).
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry.

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions.
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777).
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800).

Specimen Minimum Volume:

1 Blood spot

Forms:

1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
 - Informed Consent for Genetic Testing (T576)
 - Informed Consent for Genetic Testing-Spanish (T826)
2. Biochemical Genetics Patient Information (T602)
3. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Specimen Type	Temperature	Time	Special Container
Whole blood	Refrigerated (preferred)	90 days	
	Frozen	90 days	
	Ambient	28 days	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
PD2T	BG700	Birth Weight (grams, XXXX)	Plain Text	Yes
PD2T	BG701	Time of Birth (24hr time, XX:XX)	Plain Text	Yes
PD2T	BG702	Gestational Age (weeks, XX.X)	Plain Text	Yes

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
48436	Interpretation	Alphanumeric		63416-2
48435	Reviewed By	Alphanumeric		18771-6
BG700	Birth Weight (grams, XXXX)	Alphanumeric		8339-4
BG701	Time of Birth (24hr time, XX:XX)	Alphanumeric		57715-5
BG702	Gestational Age (weeks, XX.X)	Alphanumeric		76516-4

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

83789

Reference Values:

An interpretive report will be provided.