

Reporting Title: Pompe Disease, BS

Performing Location: Rochester

Ordering Guidance:

Due to reference range differences, this is the appropriate test for patients older than 6 weeks of age. For patients 6 weeks of age or younger, order PD2T / Pompe Disease Second-Tier Newborn Screening, Blood Spot.

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 dried on filter paper

Specimen Volume: 3 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
2. Completely fill at least 3 circles on the filter paper card (approximately 100 microliters blood per circle).
3. Let blood dry completely on the filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
4. Do not expose specimen to heat or direct sunlight.
5. Do not stack wet specimens.
6. 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)

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	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
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602290	Interpretation	Alphanumeric		59462-2
602300	Reviewed By	Alphanumeric		18771-6

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.