

Reporting Title: Beta-Glucosidase, Leukocytes**Performing Location:** Rochester**Ordering Guidance:**

This test is preferred for diagnostic testing but does not reliably detect carriers. For carrier detection, order GBAZ / Gaucher Disease, Full Gene Analysis, Varies or CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies (specify GBA Gene List ID IEMCP-M4F13T). Call 800-533-1710 to discuss testing options.

Shipping Instructions:

For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerated within 6 days of collection to be stabilized. Collect specimen Monday through Thursday only and not the day before a holiday. Specimen should be collected and packaged as close to shipping time as possible.

Specimen Requirements:

Container/Tube:

Preferred: Yellow top (ACD solution B)

Acceptable: Yellow top (ACD solution A) or lavender top (EDTA)

Specimen Volume: 6 mL

Collection Instructions: Send specimen in whole blood original tube. Do not aliquot.

Specimen Minimum Volume:

2 mL

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 ACD | Refrigerated (preferred) | 6 days | |
| | Ambient | 6 days | |

Result Codes:

| Result ID | Reporting Name | Type | Unit | LOINC® |
|-----------|------------------------------|--------------|----------------|---------|
| 606273 | Beta-Glucosidase, Leukocytes | Numeric | nmol/h/mg Prot | 32540-7 |
| 606274 | Interpretation | Alphanumeric | | 59462-2 |
| 606275 | Reviewed By | Alphanumeric | | 18771-6 |

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82963

Reference Values:

> or =3.53 nmol/hour/mg protein

An interpretative report will be provided.

Note: Results from this assay do not reflect carrier status because of individual variation of beta-glucosidase enzyme levels.