

Reporting Title: Lysosomal (Six) Panel, WBC**Performing Location:** Rochester**Ordering Guidance:**

Carrier detection using enzyme levels is unreliable for female patients with Fabry disease as results may be within the normal values. Order FABRZ / Fabry Disease, Full Gene Analysis, Varies for testing carrier status.

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 whole blood specimen in 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® |
|-----------|---------------------------------|--------------|----------------|---------|
| 606172 | Beta-Glucosidase | Numeric | nmol/h/mg Prot | 32540-7 |
| 606173 | Acid Sphingomyelinase | Numeric | nmol/h/mg Prot | 24101-8 |
| 606178 | Acid Alpha-Glucosidase | Alphanumeric | nmol/h/mg Prot | 24051-5 |
| 606179 | Acid Alpha-Glucosidase (Reflex) | Alphanumeric | nmol/h/mg Prot | 94488-4 |
| 606174 | Galactocerebrosidase | Alphanumeric | nmol/h/mg Prot | 24084-6 |
| 606175 | Galactocerebrosidase (Reflex) | Alphanumeric | nmol/h/mg Prot | 94487-6 |
| 606176 | Alpha-L-Iduronidase | Numeric | nmol/h/mg Prot | 24057-2 |
| 606177 | Alpha-Galactosidase | Numeric | nmol/h/mg Prot | 24049-9 |
| 606180 | Interpretation | Alphanumeric | | 59462-2 |
| 606181 | Reviewed By | Alphanumeric | | 18771-6 |

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657
82963
83789 (if appropriate for government payers)
82542 (if appropriate)

Reflex Tests:

| Test ID | Reporting Name | CPT Units | CPT Code | Always Performed | Orderable Separately |
|---------|------------------------------------|-----------|----------|------------------|----------------------|
| GAAWR | Acid Alpha-Glucosidase Reflex, WBC | | | No | Yes (Order GAAW) |
| GALCR | Galactocerebrosidase Reflex, WBC | | | No | Yes (Order GALCW) |

Reference Values:

Beta-Glucosidase: $> \text{ or } = 3.53 \text{ nmol/hour/mg protein}$
Acid Sphingomyelinase: $> \text{ or } = 0.32 \text{ nmol/hour/mg protein}$
Acid Alpha-Glucosidase: $> \text{ or } = 5.00 \text{ nmol/hour/mg protein}$
Galactocerebrosidase: $> \text{ or } = 1.88 \text{ nmol/hour/mg protein}$
Alpha-Galactosidase: $> \text{ or } = 10.32 \text{ nmol/hour/mg protein}$
Alpha-L-Iduronidase: $> \text{ or } = 2.06 \text{ nmol/hour/mg protein}$
Acid Alpha-Glucosidase (Reflex): $> \text{ or } = 1.50 \text{ nmol/hour/mg protein}$
Galactocerebrosidase (Reflex): $> \text{ or } = 0.300 \text{ nmol/hour/mg protein}$

An interpretative report will be provided.