

Reporting Title: Galactocerebrosidase, WBC**Performing Location:** Rochester**Ordering Guidance:**

This test will not detect carrier status. For differentiating alterations from disease-causing variants in affected patients and for carrier detection in family members, molecular sequencing of the GALC gene is necessary. Order KRABZ / Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion, Varies.

Shipping Instructions:

For optimal isolation of leukocytes, it is recommended the specimen arrive refrigerate 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®
606270	Galactocerebrosidase, WBC	Numeric	nmol/h/mg Prot	24084-6
606271	Interpretation	Alphanumeric		59462-2
606272	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82657

Reference Values:

> or =0.300 nmol/hour/mg protein
An interpretative report will be provided.