

Galactocerebrosidase, Leukocytes

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.

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 <u>Biochemical Genetics Test Request</u> (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	Туре	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



Test Definition: GALCW

Galactocerebrosidase, Leukocytes

CPT Code Information:

82657

Reference Values:

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