

## **Test Definition: LSD6W**

Lysosomal Storage Disorders, Six-Enzyme Panel, Leukocytes

# **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.

#### 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®
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



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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	Available Separately
GAAWR	Acid Alpha-Glucosidase Reflex, WBC	1	82542	No	Yes, (Order GAAW)
GALCR	Galactocerebrosidase Reflex, WBC	1	82542	No	Yes, (Order GALCW)

#### **Reference Values:**

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

An interpretative report will be provided.