

Reporting Title: Hepatosplenomegaly Panel, B**Performing Location:** Rochester**Ordering Guidance:**

This test should not be used for monitoring patients with confirmed diagnoses. If the testing requested is for monitoring purposes, see:

-CTXWB / Cerebrotendinous Xanthomatosis, Blood

-GPSYW / Glucopsychosine, Blood

-OXYWB / Oxysterols, Blood

This test's clinical sensitivity and specificity for the identification of Niemann-Pick type C (NPC) is 75% and 89%, respectively. If NPC is strongly suspected, the recommended test is HSMP / Hepatosplenomegaly Panel, Plasma.

Specimen Requirements:

Collection Container/Tube:

Preferred: Lavender top (EDTA)

Acceptable: Green top (sodium heparin, lithium heparin), yellow top (ACD B)

Specimen Volume: 1 mL

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

Specimen Minimum Volume:

0.25 mL

Forms:

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	Refrigerated (preferred)	72 hours	
	Ambient	48 hours	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
601534	Interpretation (HSMWB)	Alphanumeric		59462-2
601528	Cholestane-3beta,5alpha,6beta-triol	Numeric	nmol/mL	92756-6

Result ID	Reporting Name	Type	Unit	LOINC®
601529	Lyso-sphingomyelin	Numeric	nmol/mL	92748-3
601530	Glucopsychosine	Numeric	nmol/mL	92751-7
601531	7a-hydroxy-4-cholesten-3-one	Numeric	nmol/mL	92762-4
601532	7a,12a-dihydroxycholest-4-en-3-one	Numeric	nmol/mL	92759-0
601533	Globotriaosylsphingosine	Numeric	nmol/mL	92753-3
601535	Reviewed By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82542

Reference Values:

CHOLESTANE-3-BETA,5-ALPHA,6-BETA-TRIOL

Cutoff: < or =0.800 nmol/mL

LYSO-SPHINGOMYELIN

Cutoff: < or =0.100 nmol/mL

GLUCOPSYCHOSINE

Cutoff: < or =0.040 nmol/mL

7-ALPHA-HYDROXY-4-CHOLESTEN-3-ONE (7aC4)

Cutoff: < or =0.750 nmol/mL

7-ALPHA,12-ALPHA-DIHYDROXYCHOLEST-4-en-3-ONE (12aC4)

Cutoff: < or =0.250 nmol/mL

GLOBOTRIAOSYLSPHINGOSINE

Cutoff: < or =0.034 nmol/mL