

Reporting Title: Hepatosplenomegaly Panel, BS
Performing Location: Rochester

Ordering Guidance:
This test **should not be used** for monitoring of patients with confirmed diagnoses. If a physician is requesting testing for monitoring purposes, see:
-CTXBS / Cerebrotendinous Xanthomatosis, Blood Spot
-GPSY / Glucopsychosine, Blood Spot
-OXYBS / Oxysterols, Blood Spot

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:

- Supplies:**
-Card-Blood Spot Collection (Filter Paper) (T493)
-Card-Postmortem Screening (Filter Paper) (T525)

Container/Tube:
Preferred: Blood Spot Collection card (Filter Paper)

Acceptable: Whatman Protein Saver 903 filter paper, PerkinElmer 226 filter paper, Munktell filter paper, Postmortem Screening Card, or collected with EDTA, sodium heparin, lithium heparin, or ACD B-containing devices

Specimen Volume: 2 Blood spots

- Collection Instructions:**
- Let blood dry completely on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
 - At least 1 spot should be complete (ie, unpunched).
 - Do not expose specimen to heat or direct sunlight.
 - Do not stack wet specimens.
 - Keep specimen dry.

- Additional Information:**
- For collection instructions, see [Blood Spot Collection Instructions](#).
 - For collection instructions in Spanish, see [Blood Spot Collection Card-Spanish Instructions](#) (T777).
 - For collection instructions in Chinese, see [Blood Spot Collection Card-Chinese Instructions](#) (T800).

- Forms:**
- [1. Biochemical Genetics Patient Information](#) (T602)
 - 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)	10 days	FILTER PAPER
	Frozen	59 days	FILTER PAPER
	Ambient	10 days	FILTER PAPER

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
601526	Interpretation (HSMBS)	Alphanumeric		59462-2
601520	Cholestane-3beta,5alpha,6beta-triol	Numeric	nmol/mL	92757-4
601521	Lyso-sphingomyelin	Numeric	nmol/mL	92749-1
601522	Glucopsychosine	Numeric	nmol/mL	92752-5
601523	7a-hydroxy-4-cholesten-3-one	Numeric	nmol/mL	92763-2
601524	7a,12a-dihydroxycholest-4-en-3-one	Numeric	nmol/mL	92760-8
601525	Globotriaosylsphingosine	Numeric	nmol/mL	92754-1
601527	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 (7a-C4)

Cutoff: < or =0.750 nmol/mL

7-ALPHA,12-ALPHA-DIHYDROXYCHOLEST-4-en-3-ONE (7a12aC4)

Cutoff: < or =0.250 nmol/mL

GLOBOTRIAOSYLSPHINGOSINE

Cutoff: < or =0.034 nmol/mL