
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:

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

Additional Information:

1. For collection instructions, see Blood Spot Collection Instructions.
2. For collection instructions in Spanish, see Blood Spot Collection Card-Spanish Instructions (T777).
3. For collection instructions in Chinese, see Blood Spot Collection Card-Chinese Instructions (T800).

Specimen Minimum Volume:

1 Blood spot

Forms:

1. Biochemical Genetics Patient Information (T602)
2. 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