

Test Definition: OXYBS

Oxysterols, Blood Spot

Reporting Title: Oxysterols, BS Performing Location: Rochester

Ordering Guidance:

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 OXNP / Oxysterols, Plasma.

This test is also available as a part of a panel; see HSMBS / Hepatosplenomegaly Panel, Blood Spot. If this test (OXYBS) is ordered with either GPSY / Glucopsychosine, Blood Spot or CTXBS / Cerebrotendinous Xanthomatosis, Blood Spot, the individual tests will be canceled and HSMBS ordered.

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 the 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 <u>Blood Spot Collection Instructions</u>.
- 2. For collection instructions in Spanish, see <u>Blood Spot Collection Card-Spanish Instructions</u> (T777).
- 3. For collection instructions in Chinese, see <u>Blood Spot Collection Card-Chinese Instructions</u> (T800).

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)	10 days	FILTER PAPER
	Frozen	59 days	FILTER PAPER
	Ambient	10 days	FILTER PAPER

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
36760	Cholestane-3beta,5alpha,6beta-triol	Numeric	nmol/mL	92757-4
36761	Lyso-sphingomyelin	Numeric	nmol/mL	92749-1



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36762	Interpretation (OXYBS)	Alphanumeric	59462-2
36763	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