

## FILE DEFINITION TEST CHANGE

Notification Date: December 31, 2024 Effective Date: February 4, 2025

# Hepatosplenomegaly Panel, Blood Spot

## Test ID: HSMBS

**Explanation:** On the effective date, the Globotriaosylsphingosine analyte will be removed from the assay. Globotriaosylsphingosine is a biomarker for Fabry disease and hepatosplenomegaly is not a clinical finding in Fabry disease.

#### Current Reference Value

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-ALPAH-DIHYDROXYCHOLEST-4-en-3-ONE (12aC4) Cutoff: < or =0.250 nmol/mL

GLOBOTRIAOSYLSPHINGOSINE Cutoff: < or =0.034 nmol/mL

Current Result Codes		
Result ID	Reporting Name	
601526	Interpretation (HSMBS)	
601520	Cholestane-3beta,5alpha,6beta-triol	
601521	Lyso-sphingomyelin	
601522	Glucopsychosine	
601523	7a-hydroxy-4-cholesten-3-one	
601524	7a,12a-dihydroxycholest-4-en-3-one	
601525	Globotriaosylsphingosine	
601527	Reviewed By	

New Result Codes		
Result ID	Reporting Name	
601526	Interpretation (HSMBS)	
601520	Cholestane-3beta,5alpha,6beta-triol	
601521	Lyso-sphingomyelin	
601522	Glucopsychosine	
601523	7a-hydroxy-4-cholesten-3-one	
601524	7a,12a-dihydroxycholest-4-en-3-one	
601527	Reviewed By	

### Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.

#### New Reference Value

CHOLESTANE-3-BETA,5-ALPHA,6-BETA-TRIOL Cutoff: < or =0.800 nmol/mL

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