

Patient ID SA00173265	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Sex F	Age 42
Order Number SA00173265	Client Order Number SA00173265	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 11 Dec 2024 06:00		

Hepatosplenomegaly Panel, B

Interpretation (HSMWB)

1 MCR

POSITIVE In this sample, the concentrations of 7alpha-hydroxy-4-cholesten-3-one (7a-C4) and 7alpha,12alpha-dihydroxycholest-4-en-3-one (7a12aC4) are consistent with a biochemical diagnosis of Cerebrotendinous Xanthomatosis (CTX). Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)

Cholestane-3beta,5alpha,6beta-triol

MCR

0.800 nmol/mL

Reference Value
≤0.800

Lyso-sphingomyelin

MCR

0.100 nmol/mL

Reference Value
≤0.100

Glucopsychosine

MCR

0.040 nmol/mL

Reference Value
≤0.040

7a-hydroxy-4-cholesten-3-one

MCR

1.235 nmol/mL

Reference Value
≤0.750



7a,12a-dihydroxycholest-4-en-3-one

MCR

0.489 nmol/mL

Reference Value
≤0.250



Reviewed By

MCR

Amy Breitlow

Received: 12 Dec 2024 06:49

Reported: 12 Dec 2024 07:01

Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292