

Patient ID 321	Patient Name IMPLEMENTATION, TEST T	Birth Date 1973-05-01	Sex F	Age 51
Order Number X100485521	Client Order Number X100485521	Ordering Physician 123	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 11 Dec 2024 01:45		

Hepatosplenomegaly Panel, BS

Interpretation (HSMBS)

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

0.800 nmol/mL

MCR

Reference Value
≤0.800

Lyso-sphingomyelin

0.100 nmol/mL

MCR

Reference Value
≤0.100

Glucopsychosine

0.040 nmol/mL

MCR

Reference Value
≤0.040

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



1.235 nmol/mL

MCR

Reference Value
≤0.750

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



0.489 nmol/mL

MCR

Reference Value
≤0.250

Reviewed By

Amy Breitlow

MCR

Received: 11 Dec 2024 14:04

Reported: 11 Dec 2024 14:10

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