

Patient ID SA00142950	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 38
Order Number SA00142950	Client Order Number SA00142950	Ordering Physician CLIENT,CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 03 Mar 2021 07:00		

Sterols, P

Interpretation

1 MCR

POSITIVE In this sample, the concentration of desmosterol was elevated. Desmosterol can be elevated in both genetic (desmosterolosis/3-beta hydroxysterol delta 24 reductase deficiency) and non-genetic conditions, including amiodarone intake. If clinically indicated (multiple malformations often resembling Smith-Lemli-Opitz syndrome), consider follow-up investigations to include molecular genetic analysis of the DHCR24 gene.

Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Gas Chromatography-Mass Spectrometry (GC/MS)

7-Dehydrocholesterol

2.0 mg/L

MCR

Reference Value
≤2.0

8-Dehydrocholesterol

0.3 mg/L

MCR

Reference Value
≤0.3

8(9)-Cholestenol

5.0 mg/L

MCR

Reference Value
≤5.0

Campesterol

8.0 mg/L

MCR

Reference Value
≤8.0

Cholestanol

6.0 mg/L

MCR

Reference Value
≤6.0

Desmosterol

 34.6 mg/L

MCR

Reference Value
≤2.5

DiHydro T-MAS

0.3 mg/L

MCR

Reference Value
≤0.3

Lathosterol

6.0 mg/L

MCR

Reference Value
≤6.0

Sitosterol

15.0 mg/L

MCR

Reference Value
≤15.0

Squalene

1.0 mg/L

MCR

Reference Value
≤1.0

Stigmasterol

0.5 mg/L

MCR

Reference Value
≤0.5

Reviewed By

Amy Breitlow

MCR

Received: 04 Mar 2021 06:58

Reported: 04 Mar 2021 07:13

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	William G. Morice M.D. Ph.D	24D0404292