

Patient ID SA00114256	Patient Name TESTINGRNV, SAMPLEREPORT	Birth Date 1982-06-29	Gender F	Age 36
Order Number SA00114256	Client Order Number SA00114256	Ordering Physician CLIENT, CLIENT	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 11 Nov 2018 12:00		

CDG, S

Interpretation

1 MCR

Normal result. This finding is not consistent with a congenital disorder of glycosylation (CDG), particularly not the deficiencies of phosphomannomutase (PMM; CDG type Ia) and phosphomannoisomerase (PMI; CDG Ib). Please note that some newborn patients with these disorders intermittently normalize their transferrin glycoform pattern and may escape diagnosis. If the patient's clinical presentation is highly suggestive of a CDG, consider sending another sample to repeat the analysis in 6 to 10 weeks.

ADDITIONAL INFORMATION

Affinity Chromatography-Mass Spectrometry (MS)

Reason for Referral

MCR

CDG

Mono-oligo/Di-oligo Ratio

MCR

0.05

Reference Value
≤0.06

A-oligo/Di-oligo Ratio

MCR

0.005

Reference Value
≤0.011

Tri-sialo/Di-oligo Ratio

MCR

0.00

Reference Value
≤0.05

Apo CIII-1/Apo CIII-2 Ratio

MCR

1.45

Reference Value
≤2.91

Apo CIII-0/Apo CIII-2 Ratio

MCR

0.11

Reference Value
≤0.48

Reviewed By

MCR

Amy Breitlow

Received: 12 Nov 2018 12:51

Reported: 12 Nov 2018 12:57

CDGN, S

Interpretation

1 MCR

POSITIVE In this sample, the transferrin profile is abnormal and the N-glycan analysis is normal. These results are suggestive of one of the congenital disorders of glycosylation (CDG) type I. If clinically indicated, consider a molecular genetic panel that includes CDG type I genes. Please note that abnormal transferrin isoforms have also been observed in patients with severe liver disease of both genetic and non-genetic etiologies. Please contact the Biochemical Genetics consultant or genetic counselor on call (1-800-533-1710) if you have any questions.

ADDITIONAL INFORMATION

Matrix-assisted laser desorption ionization time-of-flight mass spectrometry (MALDI-TOF MS)

Reason for Referral

MCR

CDG

Reviewed By

MCR

Amy Breitlow

Received: 12 Nov 2018 12:51

Reported: 12 Nov 2018 13:24

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