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|---|--|--|--------------------|-----------------|
| Patient ID SA00117678 | Patient Name TESTINGRNV, SAMPLEREPOR | Birth Date 2014-06-29 | Gender F | Age 4 |
| Order Number SA00117678 | Client Order Number SA00117678 | Ordering Physician CLIENT,CLIENT | Report Notes | |
| Account Information C7028846 DLMP Rochester | | Collected 12 Feb 2019 12:00 | | |

CDG, S

Interpretation

1 MCR

Abnormal result. Although the mono-oligosaccharide transferrin to di-oligosaccharide transferrin ratios were normal, unusual transferrin glycoform variants, suggesting a transferrin isoforms missing one sialic acid; two sialic acids, and missing one sialic acid plus galactose were detected in increased amounts. Apolipoprotein-CIII profile with increased Apo CIII-1/Apo CIII-2 Ratio. These findings may be indicative of a diagnosis of CDG type II (deficiencies in subunits of the Conserved Oligomeric Golgi (COG) complex (see Zeevaert, R. Mol Genet Metab 93 (2008) 15–21). We need clinical information to provide further interpretation. Please contact the Biochemical Genetics consultant on call (Tel. 1–800–533–1710) if you have any questions or to discuss further options for diagnostic work-up.

ADDITIONAL INFORMATION

Affinity Chromatography-Mass Spectrometry (MS)

Tri-sialo/Di-oligo Ratio

 **0.13**
High

MCR

Reference Value
≤0.05

Apo CIII-1/Apo CIII-2 Ratio

 **3.75**
High

MCR

Reference Value
≤2.91

Reason for Referral

CDG

MCR

Mono-oligo/Di-oligo Ratio

0.05

MCR

Reference Value
≤0.06

A-oligo/Di-oligo Ratio

0.010

MCR

Reference Value
≤0.011

Apo CIII-0/Apo CIII-2 Ratio

0.42

MCR

Reference Value
≤0.48

Reviewed By

Amy Breitlow

MCR

Received: 13 Feb 2019 12:11

Reported: 13 Feb 2019 12:51

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 |
|------|--|--|-----------------------------|------------------|
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