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|---|--|---|--------------------|-----------------|
| Patient ID SA00050124 | Patient Name SAMPLEREPOR, CDG | Birth Date 2007-08-13 | Gender F | Age 5 |
| Order Number SA00050124 | Client Order Number SA00050124 | Ordering Physician Client, Client | Report Notes | |
| Account Information C7028846 DLMP Rochester | | Collected 28 Oct 2012 13:00 | | |

CDG, S

Reason for Referral

cdg

MCR

Interpretation

MCR

Mono-oligo/Di-oligo Ratio

0.05

MCR

Reference Value
≤0.06

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.

A-oligo/Di-oligo Ratio

0.010

MCR

Reference Value
≤0.011

ADDITIONAL INFORMATION

Affinity Chromatography-Mass Spectrometry (MS)

Tri-sialo/Di-oligo Ratio

 0.13

High

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Reference Value
≤0.05

Apo CIII-1/Apo CIII-2 Ratio

 3.75

High

MCR

Reference Value
≤2.91

Reviewed By

MCR

Jennifer Peterson

Received: 29 Oct 2012 09:22

Reported: 29 Oct 2012 09:34

Apo CIII-0/Apo CIII-2 Ratio

0.42

MCR

Reference Value
≤0.48

Performing Site Legend

| Code | Laboratory | Address |
|------|--|--|
| MCR | Mayo Clinic Dept. of Lab Med and Pathology | 200 First Street SW, Rochester, MN 55905 |