

Reporting Title: CDG, S **Performing Location:** Rochester

Ordering Guidance:

This test is for congenital disorders of glycosylation. If the ordering provider is looking for evaluation of alcohol abuse, order CDTA / Carbohydrate Deficient Transferrin, Adult, Serum.

If either PMM2-CDG (CDG-Ia) or MPI-CDG (CDG-Ib) is suspected, order PMMIL / Phosphomannomutase and Phosphomannose Isomerase, Leukocytes.

Necessary Information:

1. Patient's age is required.

2. Reason for testing is required.

Specimen Requirements:

Collection Container/Tube: Preferred: Serum gel Acceptable: Red top Submission Container/Tube: Plastic vial Specimen Volume: 0.1 mL Collection Instructions: Centrifuge and aliquot serum into a plastic vial.

Specimen Minimum Volume:

0.05 mL

Forms:

Congenital Disorders of Glycosylation (CDG, CDGN, OLIGU) Patient Information
If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

| Specimen Type | Temperature | Time | Special Container |
|---------------|--------------------|---------|-------------------|
| Serum | Frozen (preferred) | 45 days | |
| | Refrigerated | 28 days | |
| | Ambient | 7 days | |



Ask at Order Entry (AOE) Questions:

| Test ID | Question ID | Description | Туре | Reportable |
|---------|-------------|--|-------------|------------|
| CDG | BG160 | Reason for Referral: • Developmentally delayed • Congenital disorders of glycosylation • Follow-up of known patient with CDG • Evaluation of alcohol abuse - change test to 82425 (CDTA) Carb Def Transferrin, Adult, S | Answer List | Yes |

Result Codes:

| Result ID | Reporting Name | Туре | Unit | LOINC® |
|-----------|-----------------------------|--------------|------|---------|
| BG160 | Reason for Referral | Alphanumeric | | 42349-1 |
| 31721 | Mono-oligo/Di-oligo Ratio | Alphanumeric | | 35469-6 |
| 31720 | A-oligo/Di-oligo Ratio | Alphanumeric | | 35475-3 |
| 34474 | Tri-sialo/Di-oligo Ratio | Alphanumeric | | 90420-1 |
| 34476 | Apo CIII-1/Apo CIII-2 Ratio | Alphanumeric | | 90421-9 |
| 34475 | Apo CIII-0/Apo CIII-2 Ratio | Alphanumeric | | 90419-3 |
| 50820 | Interpretation | Alphanumeric | | 53808-2 |
| 50822 | Reviewed By | Alphanumeric | | 18771-6 |

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

82373

Reference Values:

| Ratio | Normal | Indeterminate | Abnormal |
|---------------------------------------|-------------|---------------|-------------|
| Transferrin mono-oligo/di-oligo ratio | < or =0.06 | 0.07-0.09 | > or =0.10 |
| Transferrin A-oligo/di-oligo ratio | < or =0.011 | 0.012-0.021 | > or =0.022 |



Test Definition: CDG

Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum

| Transferrin tri-sialo/di-oligo ratio | < or =0.05 | 0.06-0.12 | > or =0.13 |
|--------------------------------------|------------|-----------|------------|
| Apo CIII-1/Apo CIII-2 ratio | < or =2.91 | 2.92-3.68 | > or =3.69 |
| Apo CIII-0/Apo CIII-2 ratio | < or =0.48 | 0.49-0.68 | > or =0.69 |