Overview

Useful For
Screening for N-linked congenital disorders of glycosylation

Providing information on specific structural oligosaccharide abnormalities to potentially direct further genetic testing

Genetics Test Information
Testing includes matrix-assisted laser desorption/ionization time-of-flight mass spectrometry (MALDI TOF MS) analysis of N-linked oligosaccharides along with transferrin and apolipoprotein CIII isoform analysis (see CDG / Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum).

Highlights
Congenital disorders of glycosylation (CDG) comprise a large group of inborn errors of metabolism affecting predominantly N- and O-glycosylation of proteins.

N-linked CDG commonly present as clinical syndromes with multisystemic involvement and a broad clinical spectrum.

In addition to transferrin and apolipoprotein CIII isoform analysis, this test also detects and analyzes N-linked oligosaccharides by matrix-assisted laser desorption/ionization time-of-flight mass spectrometry (MALDI TOF MS) for a more comprehensive evaluation of CDG.

Additional Tests

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Reporting Name</th>
<th>Available Separately</th>
<th>Always Performed</th>
</tr>
</thead>
<tbody>
<tr>
<td>CDG</td>
<td>CDG, S</td>
<td>Yes</td>
<td>Yes</td>
</tr>
</tbody>
</table>

Testing Algorithm
When this test is ordered, carbohydrate deficient transferrin for congenital disorders will always be performed at an additional charge.

Special Instructions
- Biochemical Genetics Patient Information

Method Name
Matrix-Assisted Laser Desorption/Ionization Time-of-Flight Mass Spectrometry (MALDI TOF MS)

NY State Available
Yes

Specimen

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

Necessary Information
1. Patient’s age is required.
2. Reason for referral is required.

Specimen Required
Collection Container/Tube:
Preferred: Red Top
Acceptable: Serum gel

Submission Container/Tube: Plastic vial

Specimen Volume: 0.15 mL

Forms
1. Biochemical Genetics Patient Information (T602) in Special Instructions

Specimen Minimum Volume
0.1 mL

Reject Due To

<table>
<thead>
<tr>
<th>Gross hemolysis</th>
<th>Reject</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gross lipemia</td>
<td>OK</td>
</tr>
<tr>
<td>Gross icterus</td>
<td>OK</td>
</tr>
</tbody>
</table>

Specimen Stability Information

<table>
<thead>
<tr>
<th>Specimen Type</th>
<th>Temperature</th>
<th>Time</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serum</td>
<td>Refrigerated (preferred)</td>
<td>91 days</td>
</tr>
<tr>
<td></td>
<td>Ambient</td>
<td>91 days</td>
</tr>
<tr>
<td></td>
<td>Frozen</td>
<td>91 days</td>
</tr>
</tbody>
</table>

Clinical and Interpretive

Clinical Information
Congenital disorders of glycosylation (CDG) are a group of over 100 inherited metabolic disorders affecting largely N- and O-glycosylation of proteins. Almost 50 inborn errors of metabolism are attributed to congenital defects in N-glycosylation which takes place primarily in the cytoplasm and in the membranes of the endoplasmic reticulum. O-glycosylation defects are commonly tissue specific and present differently than classic N-linked defects. CDG are
CDGN is currently classified into 2 main groups. Type I CDG is characterized by defects in the assembly or transfer of the dolichol-linked glycan (sugar chain), while type II involves processing defects of the glycan. Depending on the specific defect, a N-glycosylation disorder can be either a type I or type II CDG.

N-linked CDG are phenotypically diverse, commonly presenting as clinical syndromes with multisystemic involvement and a broad clinical spectrum. There is considerable variation in the severity of this group of diseases ranging from a mild presentation in adults to severe multi-organ dysfunction causing infantile lethality. In some subtypes, phosphomannose isomerase (MPI)-CDG (CDG-Ib) in particular, intelligence is not compromised. CDG should be considered in all patients with multisystem disease and in those with neurological abnormalities including developmental delay and seizures, brain abnormalities such as cerebellar atrophy or hypoplasia as well as unexplained liver dysfunction. Additional common symptoms that may or may not be present include abnormal subcutaneous fat distribution, gastrointestinal issues such as vomiting, chronic diarrhea, and protein-losing enteropathy, eye abnormalities including retinal degeneration and strabismus, and cardiomyopathy.

Matrix-assisted laser desorption/ionization time-of-flight (MALDI TOF) analysis of released N-linked oligosaccharides, as is performed in this assay, is a global assessment of N-linked glycosylation. This complements the also performed transferrin and apolipoprotein CIII isoform analysis (see CDG / Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum) by providing additional information on specific structural oligosaccharide abnormalities that can in turn guide molecular testing.

**Reference Values**

Interpretative comment only.

**Interpretation**

The results of the transferrin and apolipoprotein CIII isoform analysis are followed up with matrix-assisted laser desorption/ionization time-of-flight (MALDI TOF) analysis of released N-linked oligosaccharides to assess N-linked glycosylation. Reports of abnormal results will include recommendations for additional biochemical and molecular genetic studies to more precisely identify the correct form of CDG. Treatment options, the name and telephone number of contacts who may provide studies at Mayo Clinic or elsewhere, and a telephone number for one of the laboratory directors (if the referring physician has additional questions) will be provided.

**Cautions**

No significant cautionary statements

**Clinical Reference**


**Performance**
Method Description
N-linked oligosaccharides are enzymatically released, purified, and then detected by matrix-assisted laser desorption/ionization time-of-flight mass spectrometry (MALDI TOF MS). (Unpublished Mayo method)

PDF Report
No

Day(s) and Time(s) Test Performed
Tuesday; 8 a.m.

Analytic Time
5 days (Not reported Saturday or Sunday)

Maximum Laboratory Time
10 days

Specimen Retention Time
1 month

Performing Laboratory Location
Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
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.

CPT Code Information
83789

LOINC® Information

<table>
<thead>
<tr>
<th>Test ID</th>
<th>Test Order Name</th>
<th>Order LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>CDGN</td>
<td>CDGN, S</td>
<td>In Process</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Result ID</th>
<th>Test Result Name</th>
<th>Result LOINC Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>602577</td>
<td>Interpretation</td>
<td>59462-2</td>
</tr>
<tr>
<td>BG712</td>
<td>Reason for Referral</td>
<td>42349-1</td>
</tr>
<tr>
<td>602576</td>
<td>Reviewed By</td>
<td>18771-6</td>
</tr>
</tbody>
</table>