Why Choose EGL?
- EGL offers the most comprehensive CDG testing and is the only lab offering both N-glycan structural and O-glycan analyses
- Oligosaccharides analyzed by a more specific methodology than others

<table>
<thead>
<tr>
<th>Test Code</th>
<th>Test Name</th>
<th>CPT®®® Code(s)</th>
</tr>
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<tbody>
<tr>
<td>BCDGP</td>
<td>Congenital Disorders of Glycosylation: Panel, Carbohydrate Deficient Transferrin Analysis and N-Glycan Profile, Plasma</td>
<td>82373 (x2), 83788 (x1), 84375 (x1)</td>
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<td>BNGLY</td>
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<td>BGGLY</td>
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</tr>
<tr>
<td>OS</td>
<td>Oligosaccharidosis and Congenital Disorders of Glycosylation: High resolution Oligosaccharide (Free Glycan) Profile, Urine</td>
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<td>Congenital Disorders of Glycosylation: Sequencing Panel</td>
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</tr>
<tr>
<td>DCDG1</td>
<td>Congenital Disorders of Glycosylation: Deletion/Duplication Panel</td>
<td>81228 (x1), 81479 (x1)</td>
</tr>
<tr>
<td>EXOME</td>
<td>Medical EmExome: Exome Sequencing, Proband Only</td>
<td>81415 (x1)</td>
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<tr>
<td>EXOMT</td>
<td>Medical EmExome: Exome Sequencing, Family Trios</td>
<td>81415 (x1), 81416 (x1)</td>
</tr>
</tbody>
</table>

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About Emory Genetics Laboratory (EGL)

EGL specializes in genetic diagnostic testing, with 45 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL offers a combined 1100 molecular genetics, biochemical genetics, and cytogenetics tests under one roof and custom testing for all medically relevant genes, for domestic and international clients.

Equally important to improving patient care through quality genetic testing is the contribution EGL makes back to the scientific and medical communities. EGL is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>7000 variants on >500 genes) and is also the only laboratory with a free online database (EmVClass), featuring a variant classification search and report request interface, which facilitates rapid interactive curation and reporting of variants.

Congenital Disorders of Glycosylation

EGL provides the most comprehensive testing available for the evaluation of congenital disorders of glycosylation (CDG) and offers complementary DNA testing to confirm all biochemical CDG test results. CDG are the result of alterations in protein and lipid glycosylation, and are genetic in origin. Glycosylation is the process whereby sugars (glycans) are linked together in a specific pattern and attached to proteins and lipids. Glycans signal proper cellular localization of proteins and lipids, and are needed for normal function and growth of all tissues and organs in the human body.

CDG phenotypes are extremely variable, with symptoms ranging from severe developmental delay and hypotonia beginning in infancy, to hypoglycemia and protein-losing enteropathy with normal development. CDG-Ib is the most common form reported, due to a deficiency in phosphomannomutase, an enzyme that converts mannose-6-phosphate to mannose-1-phosphate. CDG-Ib (phosphomannose isomerase deficiency) may be treated with the administration of oral mannose. CDG-Ib is the only form of CDG with an available treatment.

Consider CDG in all patients with:
- Failure to thrive
- Intellectual disability
- Hypotonia
- Hypoglycemia
- Cerebellar hypoplasia
- Partial thyroxine-binding globulin deficiency
- Perinatal dysmorphism
- Microcephaly
- Loose, wrinkled skin
- Abnormal fat pads
- Skeletal abnormalities
- Liver dysfunction
- A non-specific presentation
- Recurrent infections
- Thrombocytopenia
- Developmental delay
- Coagulopathy
- Hydrops fetalis
- Seizures and stroke-like episodes

The Path to a CDG Diagnosis

Consider Another Diagnosis or Consider Research Protocols

Biochemical Tests

Carbohydrate Deficient Transferrin Analysis

Methodology: Affinity Chromatography and MALDI TOF/TOF

N-Glycan Structural Analysis

Methodology: MALDI TOF/TOF

O-glycan Profile and Quantification

Methodology: MALDI TOF/TOF and LC MS/MS

Oligosaccharide and Glycan Screen, Urine

Methodology: MALDI TOF/TOF

CDG Next Generation Sequencing (NGS) Panel

EGL offers the CDG Next Generation Sequencing Panel to aid in the diagnosis of a variety of potential disorders, when a CDG is suspected. This panel is ideal if the gene mutation is unknown and therefore single-gene analysis won’t be cost-effective. The panel includes an analysis of 66 genes.

Whole Exome Sequencing

New types of CDG are still being discovered. Medical EmExome testing is recommended when: (1) genetic testing for a suspected condition has yielded no positive results; (2) traditional diagnostic approaches have proven ineffective; or (3) a cost effective alternative to whole genome testing is desirable. Exome sequencing has the ability to: (1) identify variants in genes that were not tested due to an atypical clinical presentation; (2) identify clinical cases in which variants from different genes contribute to the different phenotypes in the same patient; and (3) cost-effectively provide a plethora of genetic data.

For visuals of abnormal profiles refer to www.geneticslab.emory.edu/MetabolicProfiles.