Why Choose EGL Genetics?
- EGL Genetics 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.

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<th>Test Code</th>
<th>Test Name</th>
<th>CPT® Code(s)</th>
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<td>BCDGP</td>
<td>Congenital Disorders of Glycosylation: Panel, Carbohydrate Deficient Transferase Analysis and N-Glycan Profile, Plasma</td>
<td>82373 (x2), 83789 (x1), 84375 (x1)</td>
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<td>BNGLY</td>
<td>Congenital Disorders of Glycosylation: N-Glycan Profile, Qualitative, Plasma</td>
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**Congenital Disorders of Glycosylation**

About EGL Genetics

EGL Genetics specializes in genetic diagnostic testing, with nearly 50 years of clinical experience and board-certified laboratory directors and genetic counselors reporting out cases. EGL Genetics offers a combined 1000 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 Genetics makes back to the scientific and medical communities. EGL Genetics is one of only a few clinical diagnostic laboratories to openly share data with the NCBI freely available public database ClinVar (>35,000 variants on >1700 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 Genetics 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-la 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

The Path to a CDG Diagnosis

- 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

### Biochemical Tests

**Carbohydrate Deficient Transferrin Analysis**

Methodology: Affinity Chromatography and MALDI TOF/TOF

- This analysis is recommended as the first step for CDG screening.

**N-Glycan Structural Analysis**

Methodology: MALDI TOF/TOF

- N-glycan structural analysis complements the transferrin analysis with increased sensitivity and specificity.

**O-glycan Profile and Quantification**

Methodology: MALDI TOF/TOF

- This test is recommended to screen for mixed type CDGs and O-glycosylation disorders.

### Oligosaccharide and Glycan Screen, Urine

**Whole Exome Sequencing**

New types of CDGs are still being discovered. Medical Exome 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 more information about EGL Genetics:

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470.378.2200

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