
Test Code: BCDGP
Turnaround time: 2 weeks
CPT Codes: 82373 x1, 83789 x1, 84375 x1

Condition Description

Congenital disorders of glycosylation (CDGs) comprise a group of multisystem diseases with an extremely variable phenotype. Manifestations range from severe developmental delay and hypotonia with multiple organ system involvement beginning in infancy, to hypoglycemia and protein-losing enteropathy with normal development, or isolated failure to thrive.

Type I CDG comprises those disorders in which there are defects that affect the biosynthesis of doligol-linked oligosaccharides in the cytosol or the endoplasmic reticulum (ER), as well as defects involving the transfer of oligosaccharides onto nascent glycoproteins. Type II CDG comprises all defects of further trimming and elongation of N-linked oligosaccharides in the ER and Golgi.

A serum or plasma N-glycan profile can be used to identify most subtypes of CDG type II, combined type I and type II, and multiple glycosylation disorders, such as various of CDG complex deficiencies (Conserved Oligometric Golgi).

Indications

Manifestations of CDG range from severe developmental delay and hypotonia with multiple organ system involvement to hypoglycemia and protein-losing enteropathy with normal development. The diagnosis should be considered in all patients with failure to thrive, mental retardation, cerebellar hypoplasia, liver dysfunction, or seizures and stroke-like episodes.

Methodology

Affinity purification and LC-MS analysis. (Transferrin)

N-Glycan chains are released from SDS denaturated serum glycoproteins via PNGase F digestion, and then permethylated. The permethylated N-glycan are measured by liquid chromatography - mass spectrometry (LC-MS) with quadrupole - time of flight detection (QTOF). The structure of the glycans can be further analyzed by LC-MS (QTOF).

Detection

Comparing to normal serum or plasma, the changes in N-glycan structure monitored by LC-MS profile are used to identify the associated congenital disorders of glycosylation (CDG) in patient's serum or plasma.

Transferrin is reported as the ratio of mono-oligosaccharide / di-oligosaccharide transferrin and the α-oligosaccharide / di-oligosaccharide transferrin ratio.

Reference Range

The transferrin test reports qualitative results based on semi-quantitative measurement using a threshold (cut-off value) to discriminate between a positive and negative clinical interpretation.

Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test
Only one specimen type is required for this test: Frozen serum or frozen plasma.

Submit only 1 of the following specimen types

Type: Serum

Specimen Requirements:

In serum (red top) tube:
Draw 1-5 ml blood
Centrifuge immediately to separate serum and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Type: Plasma

Specimen Requirements:
In sodium heparin (green top):
Draw 1-5 ml blood
Centrifuge immediately to separate plasma and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

**Special Instructions**

Please include a recent clinic note, as well as copies of any testing, including molecular results.

**Related Tests**

- Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation (BCDGS)
- N-glycan Structural Analysis for CDG (BNGLY)
- Congenital Disorders of Glycosylation: O-glycan Profile and Quantification (BOGLY)
- Oligosaccharide and Glycan Screening (OS)
- Sequencing analysis of individual CDG genes is available.
- Sequencing analysis of different panels for CDG genes are also available.
- Custom diagnostic mutation analysis (KM) is available to family members if mutations are identified by targeted mutation testing or sequencing analysis.
- Prenatal testing is available to adult couples who are confirmed carriers of mutations. Please contact the laboratory genetic counselor to discuss appropriate testing prior to collecting a prenatal sample.