
**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**

MALDI-TOF/TOF
Affinity Chromatography / Mass Spectometry (MS)

**Detection**

Comparing to normal serum or plasma, the changes in N-glycan structure monitored by MALDI 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 a-oligosaccharide / di-oligosaccharide transferrin ratio.

**Reference Range**

Click [here](#) for reference ranges of N-glycan.

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**

Click [here](#) for the clinical presentation form.

**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.