Congenital Disorders of Glycosylation: Carbohydrate Deficient Transferrin Analysis, Plasma

Test Code: BCDGS
Turnaround time: 7 days - 10 days
CPT Codes: 82373 x1

Condition Description

Congenital disorders of glycosylation (CDG) are a group of genetic disorders caused by the alteration in synthesis and structure of protein and lipid glycosylation. In the past decade, over 50 genetic diseases have been identified that alter glycan synthesis, structure and ultimately the function of nearly all organ systems.

CDG type I (CDG-I) disorders result from impaired synthesis of the incomplete lipid linked oligosaccharide (LLO) and/or its attachment to the growing polypeptide chain. PMM2-CDG (CDG-Ia) is the most common form reported, due to phosphomannomutase deficiency, an enzyme that converts mannose-6-phosphate to mannose-1-phosphate. CDG-Ib (phosphomannose isomerase, MPI deficiency) is one of the treatable forms of CDG, by giving mannose orally.

CDG type II (CDG-II) includes defects in processing of N-linked and/or O-linked glycans. More than half of the CDG type II disorder will not be detect by transferrin alone. The combination of transferrin and N-glycan profile is highly recommended if a CDG type II disorder is suspected.

Phenotypes of this disorder are extremely variable. 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. However most subtypes have been described in only a few individuals, and thus understanding of the phenotypes of most CDG subtypes is limited.

Educational Materials:
- CDG Handout for Parents
- CDG Handout for Clinicians

References:

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 stroke-like episodes.

Methodology

Affinity Chromatography / Mass Spectrometry (MS) Affinity purification and LC-MS analysis

Detection

Ratio of mono-oligosaccharide / di-oligosaccharide transferrin and the a-oligosaccharide / di-oligosaccharide transferrin ratio. Note the majority of CDG type II may not be detected by this test alone. The combination of transferrin and N-glycan profiles is highly recommended to improve the detection of CDG.

Reference Range

The 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

Submit only 1 of the following specimen types

Type: Whole Blood (Sodium Heparin)

Specimen Requirements:
Sodium Heparin (Green Top)
1-2 ml

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday. (Late Friday collections may be stored at room temperature over the weekend for Monday receipt.)

Type: Whole Blood (No additive)

Specimen Requirements:
No Additive (Red Top) or SST (Serum Separator Tube - no additives)
1-2 ml

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday

Type: Plasma

Specimen Requirements:
Sodium Heparin (Green Top)
1-2 ml
Sample should be collected while fasting or 2-4 hours post prandial. Centrifuge to separate plasma and freeze.

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

Type: Serum

Specimen Requirements:
Clean container without additives
1-2 ml
Spin down, transfer, and ship frozen.

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

Special Instructions

Click here for the clinical presentation form.

Related Tests

- N-glycan Structural Analysis for CDG (BNGLY)
- N-glycan and Carbohydrate Deficient Transferrin Panel for CDG (BCDGP)
- 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.