Lab Dept: Anatomic Pathology

Test Name: N-GLYCAN STRUCTURAL ANALYSIS FOR CDG SYNDROME

General Information

Lab Order Codes: NGLYC

Synonyms: Carbohydrate Deficient Glycoprotein Syndrome; N-Glycan Profile; Congenital Disorders of Glycosylation

CPT Codes:
82373 - Carbohydrate deficient transferrin
83789 - Mass spectrometry and tandem mass spectrometry (MS, MS/MS), non-drug analyte, not elsewhere specified; qualitative or quantitative, each specimen
84375 - Sugars, chromatographic, TLC or paper chromatography

Test Includes: Most subtypes of CDG type II, combined type I and type II, and multiple glycosylation disorders, such as various types of COG complex deficiencies (Conserved Oligometric Golgi).

Logistics

Test 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.

Lab Testing Sections: Anatomic Pathology – Sendouts

Referred to: Emory Genetics Laboratory (Test code: BNGLY)

Phone Numbers:
MIN Lab: 612-813-6280
STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 7 – 10 days

Special Instructions: Please submit clinical history form with the patient or specimen presenting at the lab.
**Specimen**

Specimen Type: Blood

Container: Red top tube

Draw Volume: 1 - 5 mL (Minimum: 1 mL) blood

Processed Volume: 0.3 – 1.5 (Minimum: 0.3 mL) serum

Collection: Routine venipuncture for blood specimens

Special Processing: Lab Staff: Centrifuge immediately to separate serum and freeze. Ship frozen sample on dry ice with overnight courier. Please provide clinical information.

Patient Preparation: None

Sample Rejection: Mislabeled or unlabeled specimen

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**Interpretive**

Reference Range: Interpretive report

Critical Values: N/A

Limitations: N/A

Methodology: N-Glycan chains are firstly released from SDS denaturated serum glycoproteins via PNGase F digestion, and then permethylated. The permethylated N-glycan are measured by matrix-assisted laser desorption/ionization time of light mass spectrometry (MALDI-TOF). The structure of the glycans can be further analyzed by MALDI-TOF/TOF.

References: Emory Genetics Laboratory (February 2017)
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Tucker, GA  30084
(470) 378-2200  Fax (470) 378-2250

Updates: 1/26/2016: CPT update
2/23/2017: Emory address change