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 (Conservated 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 <u>clinical history form</u> 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
Interpretive	
Reference Range:	Interpretive report
Critical Values:	N/A
Critical Values: Limitations:	N/A N/A
Critical Values: Limitations: Methodology:	N/A N/A 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.
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