Overview

Useful For

Screening for N-linked congenital disorders of glycosylation

Providing information on specific structural oligosaccharide abnormalities to potentially direct further genetic testing

Genetics Test Information

Congenital disorders of glycosylation (CDG) comprise a large group of inborn errors of metabolism affecting predominantly N- and O-glycosylation of proteins.

N-linked CDG commonly present as clinical syndromes with multisystemic involvement and a broad clinical spectrum.

In addition to transferrin and apolipoprotein CIII isoform analysis, this test also detects and analyzes N-linked oligosaccharides by matrix-assisted laser desorption/ionization time-of-flight mass spectrometry (MALDI TOF MS) for a more comprehensive evaluation of CDG.

Additional Tests

Test ID	Reporting Name	Available Separately	Always Performed
CDG	CDG, S	Yes	Yes

Testing Algorithm

When this test is ordered, carbohydrate deficient transferrin for congenital disorders will always be performed at an additional charge.

Special Instructions

- Biochemical Genetics Patient Information

Method Name

Matrix-Assisted Laser Desorption/Ionization Time-of-Flight Mass Spectrometry (MALDI-TOF MS)

NY State Available

Yes

Specimen

Specimen Type

Serum

Advisory Information

This test is for congenital disorders of glycosylation. For evaluation of alcohol abuse, order CDTA / Carbohydrate Deficient Transferrin, Adult, Serum.

Necessary Information

1. Patientâ€™s age is required.

2. Reason for testing is required.

Specimen Required

Collection Container/Tube:Â

Preferred: Red Top

Acceptable: Serum gel

Submission Container/Tube: Plastic vial

Specimen Volume: 0.15 mL

Forms

Biochemical Genetics Patient Information (T602) in Special Instructions

Specimen Minimum Volume

0.1 mL

Reject Due To

Gross hemolysis	Reject
Gross lipemia	OK
Gross icterus	OK

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Serum	Refrigerated (preferred)	91 days	
	Ambient	91 days	
	Frozen	91 days	

Clinical and Interpretive

Clinical Information

Congenital disorders of glycosylation (CDG) are a group of over 100 inherited metabolic disorders affecting largely N- and O-glycosylation of proteins. Almost 50 inborn errors of metabolism are attributed to congenital defects in N-glycosylation, which takes place primarily in the cytoplasm and in the membranes of the endoplasmic reticulum. O-glycosylation defects are commonly tissue specific and present differently than classic N-linked defects. CDG are currently classified into 2 main groups. Type I CDG is characterized by defects in the assembly or transfer of the dolichol-linked glycan (sugar chain), while type II involves processing defects of the glycan. Depending on the specific defect, an N-glycosylation disorder can be either a type I or type II CDG.

N-linked CDG are phenotypically diverse, commonly presenting as clinical syndromes with multisystemic involvement and a broad clinical spectrum. There is considerable variation in the severity of this group of diseases ranging from a mild presentation in adults to severe multi-organ dysfunction causing infantile lethality. Intellectual disability is common, although in some subtypes, phosphomannose isomerase (MPI)-CDG (CDG-Ib) in particular,

this is not observed. CDG should be considered in all patients with multisystem disease and in those with neurologic abnormalities including developmental delay and seizures, brain abnormalities such as cerebellar atrophy or hypoplasia as well as unexplained liver dysfunction. Additional common symptoms that may or may not be present include abnormal subcutaneous fat distribution, gastrointestinal issues such as vomiting, chronic diarrhea, and protein-losing enteropathy, eye abnormalities including retinal degeneration and strabismus, and cardiomyopathy.

Matrix-assisted laser desorption/ionization time-of-flight (MALDI TOF) analysis of released N-linked oligosaccharides, as is performed in this assay, is a global assessment of N-linked glycosylation. This complements the also performed transferrin and apolipoprotein CIII isoform analysis (see CDG / Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum) by providing additional information on specific structural oligosaccharide abnormalities that can in turn guide molecular testing.

Reference Values

Interpretative comment only.

Interpretation

The results of the transferrin and apolipoprotein CIII isoform analysis are followed up with matrix-assisted laser desorption/ionization time-of-flight (MALDI TOF) analysis of released N-linked oligosaccharides to assess N-linked glycosylation. Reports of abnormal results will include recommendations for additional biochemical and molecular genetic studies to more precisely identify the specific congenital disorder of glycosylation (CDG). Treatment options, the name and telephone number of contacts who may provide studies, and a telephone number for one of the laboratory directors (if the referring physician has additional questions) will be provided.

Cautions

No significant cautionary statements

Clinical Reference

1. Freeze HH: Congenital disorders of glycosylation: CDG-I, CDG II, and beyond. Curr Mol Med. 2007;7:389-396. doi: 10.2174/156652407780831548

2. Freeze HH, Eklund EA, Ng BG, Patterson MC: Neurology of inherited glycosylation disorders. Lancet Neurol. 2012;11:453- 466. doi: 10.1016/S1474-4422(12)70040-6

3. Hennet T, Cabalzar J: Congenital disorders of glycosylation: a concise chart of glycocalyx dysfunction. Trends Biochem Sci. 2015 Jul;40(7):377-384. doi: 10.1016/j.tibs.2015.03.002

4. Freeze HH, Chong JX, Bamshad MJ, Ng BG: Solving glycosylation disorders: fundamental approaches reveal complicated pathways. Am J Hum Genet. 2014 Feb 6;94(2):161-175. doi: 10.1016/j.ajhg.2013.10.024

5. Scott K, Gadomski T, Kozicz, Morava E: Congenital disorders of glycosylation: new defects and still counting. J Inherit Metab Dis. 2014;37:609-617. doi: 10.1007/s10545-014-9720-9

Performance

Method Description

N-linked oligosaccharides are enzymatically released, purified, and then detected by matrix-assisted laser desorption/ionization time-of-flight mass spectrometry (MALDI-TOF MS).(Unpublished Mayo method)

PDF Report

No

Day(s) and Time(s) Test Performed

Tuesday; 8 a.m.

Analytic Time

5 days (Not reported Saturday or Sunday)

Maximum Laboratory Time

10 days

Specimen Retention Time

1 month

Performing Laboratory Location

Rochester

Fees and Codes

Fees

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information

83789

LOINC® Information

Test ID	Test Order Name	Order LOINC Value
CDGN	CDGN, S	In Process

Result ID	Test Result Name	Result LOINC Value
602577	Interpretation	59462-2
BG712	Reason for Referral	42349-1
602576	Reviewed By	18771-6