MAYO CLINIC LABORATORIES

Test Definition: CDGN

Congenital Disorders of N-Glycosylation, Serum

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 serum N-linked oligosaccharides by matrix-assisted laser desorption/ionization time-of-flight mass spectrometry 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.

For more information see Congenital Disorders of Glycosylation: Screening Algorithm.

Special Instructions

- Congenital Disorders of Glycosylation: Screening Algorithm
- Congenital Disorders of Glycosylation 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

Ordering Guidance

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

Necessary Information

1. Patient's age is required.

2. Reason for testing is required.

Specimen Required

Collection Container/Tube:

Preferred: Serum gel

Acceptable: Red top

Submission Container/Tube: Plastic vial

Specimen Volume: 0.15 mL

Collection Instructions: Centrifuge and aliquot serum into a plastic vial.

Forms

1. Congenital Disorders of Glycosylation Patient Information

2. If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

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)	28 days	
	Ambient	7 days	
	Frozen	45 days	

Clinical & Interpretive

Clinical Information

Congenital disorders of glycosylation (CDG) are a group of over 150 inherited metabolic disorders largely affecting N- and O-glycosylation of proteins. The majority of CDG are attributed to congenital defects in N-glycosylation, which take place primarily in the cytoplasm and in the membranes of the endoplasmic reticulum. O-glycosylation defects are frequently 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, usually 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-CDG (MPI-CDG or CDG type Ib) in particular, intellect is preserved. 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; and unexplained liver dysfunction. Additional common symptoms that may 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) mass spectrometry analysis of released N-linked oligosaccharides, as 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 guide molecular testing.

Reference Values

An interpretive report will be provided.

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) mass spectrometry 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 identify more precisely the specific congenital disorder of glycosylation. If applicable, 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. Sparks SE, Krasnewich DM. Congenital disorders of N-linked glycosylation and multiple pathway overview. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews [Internet]. University of Washington, Seattle; 2005. Updated January 12, 2017. Accessed March 1, 2024. Available at www.ncbi.nlm.nih.gov/books/NBK1332/
2. Chang IJ, He M, Lam CT. Congenital disorders of glycosylation. Ann Transl Med. 2018;6(24):477. doi:10.21037/atm.2018.10.45

3. Francisco R, Marques-da-Silva D, Brasil S, et al. The challenge of CDG diagnosis. Mol Genet Metab. 2019;126(1):1-5. doi:10.1016/j.ymgme.2018.11.003
4. Freeze HH, Chong JX, Bamshad MJ, Ng BG. Solving glycosylation disorders: fundamental approaches reveal complicated pathways. Am J Hum Genet. 2014;94(2):161-175. doi:10.1016/j.ajhg.2013.10.024
5. Verheijen J, Tahata S, Kozicz T, et al. Therapeutic approaches in congenital disorders of glycosylation (CDG) involving N-linked glycosylation: an update. Genet Med. 2020;22(2):268-279. doi:10.1038/s41436-019-0647-2
6. Francisco R, Brasil S, Poejo J, et al. Congenital disorders of glycosylation (CDG): state of the art in 2022. Orphanet J Rare Dis. 2023;18(1):329. doi:10.1186/s13023-023-02879-z

Performance

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

PDF Report
No

Day(s) Performed
Wednesday

Report Available
5 to 11 days

Specimen Retention Time
1 month

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & 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 account representative. 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. It has not been cleared or approved by the US Food and Drug Administration.

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Congenital Disorders of N-Glycosylation, Serum

CPT Code Information

83789

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CDGN	CDGN, S	90417-7

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