Congenital disorders of glycosylation (CDG) scheme
Web submission report

Laboratoire N: 412 (CDG Nijmegen)

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Date of sample received : 2023-02-13

Date of reporting results : 12 May 2023 - 19:12:04

The results below have been sent and saved in the CSCQ database at the date and time indicated above.

23-04-DGN / CDG-PP-2023-A — Lab 412

Clinical picture

Patient sex : F Age at diagnosis : 21 Year(s) Age present : 21 Year(s)

Hepatic fibrosis, increased gamma-glutamyl transferase (GGT)

1. Analytical Results

Sialotransferrin (CDG) — **CDG-PP-2023-A Lab 412**

Analyte	Method	Isoform	Result (unit)	Lower ref.	Upper ref.

Comments User Initials : USR1

Not performed

2. Interpretation

User Initials : USR1 — **Lab 412**

Interpretation of the profiles (normal or abnormal, profile type: suggestive for CDG-I, CDG-II or other)

CDG type1 abnormalities on transferrin (Trf) and Alpha-1 Anti-trypsin (AAT). Suggestive for CDG-I.

3. Recommendations

User Initials : USR1 — **Lab 412**

CDG-I profile. Secondary causes of CDG should be excluded (hereditary fructose intolerance, galactosemia, liver disease, chronic alcool abuse). Ask for EDTA blood sample to test PMM2 (PMM2-CDG) and MPI (MPI-CDG) enzymatic activities. Ask for skin fibroblasts to possibly test other enzymatic activities. EDTA blood sample and skin biopsy will also be used for molecular sequencing (CDG gene panels, WES...). Ask for an informed consent for genetic studies.

4. Files uploaded

User Initials : — **Lab 412**

File number	File name	Comment
1	EEQ ERNDIM CDG-PP-2023-A-B-C - WB Trf and AAT - Bichat Paris lab 412.pdf	
2	EEQ ERNDIM CDG-PP-2023-A-B-C - WB Trf and AAT - Bichat Paris lab 412.pdf	
3	EEQ ERNDIM CDG-PP-2023-A-B-C - WB Trf and AAT - Bichat Paris lab 412.pdf	

23-04-DGN / CDG-PP-2023-B — Lab 412

Clinical picture

Patient sex : M Age at diagnosis : 2 Year(s) Age present : 2 Year(s)

Polycystic kidney disease, hyperinsulinemic hypoglycemia

1. Analytical Results

Sialotransferrin (CDG) **CDG-PP-2023-B Lab 412**

Analyte	Method	Isoform	Result (unit)	Lower ref.	Upper ref.

Comments User Initials : USR1
Not performed

2. Interpretation

User Initials : USR1 **Lab 412**

Interpretation of the profiles (normal or abnormal, profile type: suggestive for CDG-I, CDG-II or other)
Normal profiles. Not suggestive for CDG.

3. Recommendations

User Initials : USR1 **Lab 412**

Normal profile. Do not propose anything in the field of CDG.

4. Files uploaded

No files uploaded.

23-04-DGN / CDG-PP-2023-C **Lab 412**

Clinical picture

Patient sex : F Age at diagnosis : 17 Year(s) Age present : 17 Year(s)
Mild intellectual disability, pigmentary retinopathy and slurred speech.

1. Analytical Results

Sialotransferrin (CDG) **CDG-PP-2023-C Lab 412**

Analyte	Method	Isoform	Result (unit)	Lower ref.	Upper ref.

Comments User Initials : USR1
Not performed

2. Interpretation

User Initials : USR1 **Lab 412**

Interpretation of the profiles (normal or abnormal, profile type: suggestive for CDG-I, CDG-II or other)
CDG type1 abnormalities on transferrin (Trf) and Alpha-1 Anti-trypsin (AAT). Suggestive for CDG-I.

3. Recommendations

User Initials : USR1 **Lab 412**

CDG-I profile. Secondary causes of CDG should be excluded (hereditary fructose intolerance, galactosemia, liver disease, chronic alcool abuse). Ask for EDTA blood sample to test PMM2 (PMM2-CDG) and MPI (MPI-CDG) enzymatic activities. Ask for skin fibroblasts to possibly test other enzymatic activities. EDTA blood sample and/or skin biopsy will also be used for molecular sequencing (CDG gene panels, WES...). Ask for an informed consent for genetic studies.

4. Files uploaded

No files uploaded.