# 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 : 26 September 2023 - 14:28:27

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

## 23-08-DGN / CDG-PP-2023-D — Lab 412

**Clinical picture**

Patient sex : F Age at diagnosis : 15 Year(s) Age present : 15 Year(s)

Axial hypotonia, mild-moderate intellectual disability, abnormalities in coagulation

### 1. Analytical Results

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

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

Comments User Initials : USR1

Not applicable

### 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), alpha-1 Anti-trypsin (AAT) and haptoglobin. Suggestive for CDG-I.

### 3. Recommendations

User Initials : USR1 — **Lab 412**

CDG-I profile(s). Secondary causes of CDG should be excluded (hereditary fructose intolerance, galactosemia, liver disease, chronic alcool abuse). Eventually, 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 fibroblasts will also be used for molecular sequencing (CDG gene panels, WES...). Ask for an informed consent for genetic studies.

### 4. Files uploaded

User Initials : ABR — **Lab 412**

| File number | File name | Comment |
|---|---|---|
| 1 | EEQ ERNDIM 2023-D-E-F.pdf | |

## 23-08-DGN / CDG-PP-2023-E — Lab 412

**Clinical picture**

Patient sex : F Age at diagnosis : 3 Year(s) Age present : 3 Year(s)

Seizure, axial hypotonia

### 1. Analytical Results

**Sialotransferrin (CDG)** — **CDG-PP-2023-E 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 profile(s). 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

User Initials : ABR **Lab 412**

| File number | File name | Comment |
|---|---|---|
| 1 | EEQ ERNDIM 2023-D-E-F.pdf | |

## 23-08-DGN / CDG-PP-2023-F **Lab 412**

**Clinical picture**

Patient sex : M Age at diagnosis : 4 Year(s) Age present : 4 Year(s)
Global developmental delay, autism spectrum disorder, bruising susceptibility

### 1. Analytical Results

**Sialotransferrin (CDG)** **CDG-PP-2023-F 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 profile(s). Possible transferrin protein variant? According to AAT and haptoglobin 2-DE patterns, not suggestive for CDG.

### 3. Recommendations

User Initials : USR1 **Lab 412**

Normal AAT and haptoglobin 2-DE patterns. At our side, discrete doubt on a possible transferrin variant. Although clinically not evocative, a MAN1B1-CDG could not formally be excluded. Indeed, MAN1B1-CDG classically present with an isolated increase of the 3-sialo transferrin fraction suggestive for a variant. Considering the very low volume of available sample (<25 µL), we are not able to perform neuraminidase treatment to exclude a Trf variant.

### 4. Files uploaded

User Initials : ABR **Lab 412**

| File number | File name | Comment |
|---|---|---|
| 1 | EEQ ERNDIM 2023-D-E-F.pdf | |