Congenital disorders of glycosylation (CDG) scheme Web submission report

Laboratoire N: 412 (CDG Porto)

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Date of sample received : 2025-03-24

Date of reporting results : 13 May 2025 - 10:08:45

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

25-04-DGN / CDG-PP-2025-A				Lab 412
Clinical picture Patient sex : F Age at diagnosis : 17 Year(s) Hips lipodystrophy, terminal nystagmus, without autonomou	us walking	Age present : 17	' Year(s)	
1. ANALYTICAL RESULTS Sialotransferrin (CDG)			CDG-PP-2025-/	\ lah 412
Analyte Method	Isoform	Result (unit)	Lower ref.	Upper ref.
Comments User Initials : USR1 Not performed				
2. INTERPRETATION				l ah 412
Interpretation of the profiles (normal or abnormal, profile typ Abnormal profile: suggestive for CDG-I.	be: suggestive for CI	DG-I, CDG-II or oth	ner)	
3. RECOMMENDATIONS User Initials : USR1 CDG-I profile. Secondary causes of CDG should be exclud chronic alcool abuse). Ask for EDTA blood sample and/or s CDG) enzymatic activities. EDTA blood sample and skin bir panels, WES). Ask for an informed consent for genetic st	ed (hereditary fructo kin fibroblasts to pos opsy will also be use udies.	se intolerance, gal ssibly test PMM2 (l d for molecular se	actosemia, liver o PMM2-CDG) and quencing (CDG g	Lab 412 lisease, MPI (MPI- ene
4. FILES UPLOADED User Initials :				Lab 412
File number File name		Comment		
1 ERNDIM-CDG-ABC-2025.pdf				
25-04-DGN / CDG-PP-2025-B				Lab 412
Clinical picture Patient sex : F Age at diagnosis : 4 Year(s) Inverted nipples, epilepsy, liver dysfunction		Age present: 4	Year(s)	
1. Analytical Results				
Analyte Method	Isoform	Result (unit)	Lower ref.	Lab Upper ref.
Comments User Initials : USR1 Not performed				

Lab 412

Lab 412

2. INTERPRETATION

User Initials : USR1 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

Normal profiles. Little doubt on a possible transferrin variant not related to a glycosylation defect. Considering the low volume of available sample, we are not able to perform neuraminidase treatment to exclude it. Do not propose anything in the field of CDG. Nevertheless, since falsely normal profils have been described in some CDG cases, the diagnosis cannot be totally excluded.

4. FILES UPLOADED

User Initials :				Lab 412
File number File name		Comment		
1 ERNDIM-CDG-ABC-2025.pdf				
25-04-DGN / CDG-PP-2025-C				Lab 412
Clinical picture				
Patient sex : MAge at diagnosis : 30 Year(s)Mild intellectual disability, hearing impairment, epilepsy		Age present : 30 Year(s)		
1. Analytical Results				
Analyte Method	Isoform	Result (unit)	Lower ref.	Lab Upper ref.
Comments User Initials : USR1 Not performed				
2. Interpretation				
User Initials : USR1 Interpretation of the profiles (normal or abnormal, profile typ Abnormal profile: suggestive for CDG-I.	be: suggestive for CD	DG-I, CDG-II or other	·)	Lab 412
3. Recommendations				
User Initials : USR1 CDG-I profile. Secondary causes of CDG should be exclud chronic alcool abuse). Ask for EDTA blood sample and/or s CDG) enzymatic activities. EDTA blood sample and skin bio panels, WES). Ask for an informed consent for genetic st	ed (hereditary fructos kin fibroblasts to pos opsy will also be used udies.	se intolerance, galac sibly test PMM2 (PM d for molecular sequ	tosemia, liver o IM2-CDG) and encing (CDG g	Lab 412 disease, I MPI (MPI- gene
3. RECOMMENDATIONS User Initials : USR1 CDG-I profile. Secondary causes of CDG should be exclud chronic alcool abuse). Ask for EDTA blood sample and/or s CDG) enzymatic activities. EDTA blood sample and skin bio panels, WES). Ask for an informed consent for genetic st	ed (hereditary fructos kin fibroblasts to pos opsy will also be used udies.	se intolerance, galac sibly test PMM2 (PM d for molecular sequ	tosemia, liver o IM2-CDG) and encing (CDG g	Lab 412 disease, I MPI (MPI- gene
4. FILES UPLOADED				

User Initials :			Lab 412
File number	File name	Comment	
1	ERNDIM-CDG-ABC-2025.pdf		

13 May 2025 - 10:08:46 1 © CSCQ 2019, v 1.1