

## Congenital Disorders of Glycosylation (CDG) 2023 EQA Sample Diagnoses

**ERNDIM Administration Office**  
c/o EMQN CIC, Unit 4, Enterprise House,  
Manchester Science Park, Pencroft Way,  
Manchester, M15 6SE,  
United Kingdom  
**Tel:** +44 161 757 4952  
**Fax:** +44 161 850 1145  
**Email:** [admin@erndim.org](mailto:admin@erndim.org)

**Scientific Advisor**  
Dr Dulce Quelhas  
Unidade Bioquimica Genetica  
Centro de Genetica Medica Jacinto de  
Magalhaes, Centro Hospitalar do Porto, EPE,  
Pr Pedro Nunes 88  
Porto, 4099-028, Portugal  
**Email:** [dulce.quelhas@chporto.min-saude.pt](mailto:dulce.quelhas@chporto.min-saude.pt)

**Scheme Organisers**

**1. Sample dispatch**

Dr Eline van der Hagen  
Streekziekenhuis Koningin Beatrix  
MCA Laboratory  
Beatrixpark 1  
7101 BN Winterswijk  
The Netherlands  
**Email:** [mca.office@skbwinterswijk.nl](mailto:mca.office@skbwinterswijk.nl)

**2. Results Website**

**a) Alessandro Salemma; b) Nicola Braik**  
CSCQ, Swiss Center for Quality Control  
2 chemin du Petit-Bel-Air  
CH-1225 Chêne-Bourg  
Switzerland  
**Email:** **a)** [Alessandro.Salemma@hcuge.ch](mailto:Alessandro.Salemma@hcuge.ch); **b)** [Nicola.Braik@hcuge.ch](mailto:Nicola.Braik@hcuge.ch)

Dear Participant,

Please find below the diagnoses for the 2023 CDG EQA scheme.

Sample ID	Clinical information (sex, age, phenotype)	Sex	Patient Age	Diagnoses
<b>2023-A</b>	Hepatic fibrosis, gamma-glutamyl transferase (GGT)	F	21 years	Alcohol Abuse sample
<b>2023-B</b>	Polycystic kidney disease, hyperinsulinemic hypoglycemia	M	2 years	Normal sample
<b>2023-C</b>	Mild intellectual disability, pigmentary retinopathy and slurred speech.	F	17 years	PMM2-CDG
<b>2023-D</b>	Axial hypotonia, mild-moderate intellectual disability, Abnormalities in coagulation	F	15 years	PMM2-CDG
<b>2023-E</b>	Seizure, Axial hypotonia	F	3 years	Normal serum
<b>2023-F</b>	Global developmental delay, autism spectrum disorder, bruising susceptibility	M	4 years	Transferrin variant

With regards,  
ERNDIM Administration Team

**On behalf of**  
**Dr Dulce Quelhas**  
**Scientific Advisor, CDG Scheme**